- Secondary (Central) hyperthyroidism
  - TSH-secreting pituitary adenoma
- Familial hyperthyroidism
  - Hereditary hyperthyroidism [thyrotoxicosis] due to mutations in TSH receptor
  - Selective pituitary resistance to thyroid hormone

Thyroiditis
- Riedel thyroiditis
- Acute thyroiditis
  - Acute thyroiditis due to bacterial infection
  - Acute thyroiditis due to fungal infection
- Subacute thyroiditis
- Autoimmune thyroiditis
  - Hashimoto thyroiditis
  - Painless (silent) thyroiditis
  - Postpartum thyroiditis
- Drug-induced thyroiditis
- Radiation thyroiditis after 131 I treatment
- Other chronic thyroiditis
- ~~Thyroiditis, unspecified~~

Certain specified disorders of thyroid
- Hypersecretion of calcitonin
- Generalized resistance to thyroid hormone
- ~~Other specified disorders of thyroid~~
- ~~Disorder of thyroid, unspecified~~

Diabetes mellitus and intermediate hyperglycaemia

Supplementary classification for use with certain specified diabetes mellitus
- Genetic defects of beta cell function associated with Other specified diabetes mellitus
- Genetic defects in insulin action associated with other specified diabetes mellitus
- Diseases of exocrine pancreas associated with Other specified diabetes mellitus
- Endocrinopathies associated with other specified diabetes mellitus
- Drug or chemical induced associated with Other specified diabetes mellitus
- Infections associated with Other specified diabetes mellitus
- Uncommon forms of immune-mediated diabetes mellitus associated with Other specified diabetes mellitus
- Other genetic syndromes associated with Other specified diabetes mellitus
- Other clinically defined subgroups associated with Other specified diabetes mellitus

Type 1 diabetes mellitus
- Immune-related Type 1 diabetes mellitus
  - Immune-related Type 1 diabetes mellitus with coma
    - Immune-related Type 1 diabetes mellitus with hypoglycaemic coma
    - Immune-related Type 1 diabetes mellitus with hyperosmolar hyperglycaemic coma without ketoacidosis
  - Immune-related Type 1 diabetes mellitus with acidosis
    - Immune-related Type 1 diabetes mellitus with ketoacidosis, without coma
    - Immune-related Type 1 diabetes mellitus with ketoacidosis, with coma
    - Immune-related Type 1 diabetes mellitus with lactic acidosis
    - Immune-related Type 1 diabetes mellitus with metabolic acidosis
  - Immune-related Type 1 diabetes mellitus with kidney complication
  - Immune-related Type 1 diabetes mellitus with ophthalmic complications
    - Immune-related Type 1 diabetes mellitus with retinopathy
      - Immune-related Type 1 diabetes mellitus with simple retinopathy
      - Immune-related Type 1 diabetes mellitus with preproliferative retinopathy
      - Immune-related Type 1 diabetes mellitus with proliferative retinopathy
    - Immune-related Type 1 diabetes mellitus with macular oedema
    - Immune-related Type 1 diabetes mellitus diabetic cataract
    - Immune-related Type 1 diabetes mellitus with glaucoma
  - Immune-related Type 1 diabetes mellitus with neurological complications
    - Immune-related Type 1 diabetes mellitus with mononeuropathy
    - Immune-related Type 1 diabetes mellitus with polyneuropathy
    - Immune-related Type 1 diabetes mellitus with other neurological complications
  - Immune-related Type 1 diabetes mellitus with macrovascular complications
    - Immune-related Type 1 diabetes mellitus with cerebrovascular disease
    - Immune-related Type 1 diabetes mellitus with coronary artery disease
    - Immune-related Type 1 diabetes mellitus with peripheral artery disease
  - Immune-related Type 1 diabetes mellitus with other specified complications
    - Immune-related Type 1 diabetes mellitus with diabetic foot
    - Immune-related Type 1 diabetes mellitus with diabetic skin lesions
    - Immune-related Type 1 diabetes mellitus with periodontal disease
  - Immune-related Type 1 diabetes mellitus with unspecified complications
  - Immune-related Type 1 diabetes mellitus without complications
- Idiopathic Type 1 diabetes mellitus
  - Idiopathic Type 1 diabetes mellitus with complications
    - Idiopathic Type 1 diabetes mellitus with coma
      - Idiopathic Type 1 diabetes mellitus with hypoglycaemic coma
      - Idiopathic Type 1 diabetes mellitus with hyperosmolar hyperglycaemic coma without ketoacidosis
    - Idiopathic Type 1 diabetes mellitus with acidosis
      - Idiopathic Type 1 diabetes mellitus with ketoacidosis, without coma
      - Idiopathic Type 1 diabetes mellitus with ketoacidosis, with coma
      - Idiopathic Type 1 diabetes mellitus with lactic acidosis
      - Idiopathic Type 1 diabetes mellitus with metabolic acidosis
    - Idiopathic Type 1 diabetes mellitus with kidney complications
    - Idiopathic Type 1 diabetes mellitus with ophthalmic complications
      - Idiopathic Type 1 diabetes mellitus with retinopathy
        - Idiopathic Type 1 diabetes mellitus with simple retinopathy
        - Idiopathic Type 1 diabetes mellitus with preproliferative retinopathy
        - Idiopathic Type 1 diabetes mellitus with proliferative retinopathy
      - Idiopathic Type 1 diabetes mellitus with macular oedema
      - Idiopathic Type 1 diabetes mellitus with diabetic cataract
      - Idiopathic Type 1 diabetes mellitus with glaucoma
    - Idiopathic Type 1 diabetes mellitus with neurological complications
      - Idiopathic Type 1 diabetes mellitus with mononeuropathy
      - Idiopathic Type 1 diabetes mellitus with polyneuropathy
      - Idiopathic Type 1 diabetes mellitus with other neurological complication
    - Idiopathic Type 1 diabetes mellitus with macrovascular complications
      - Idiopathic Type 1 diabetes mellitus with cerebrovascular disease
      - Idiopathic Type 1 diabetes mellitus with coronary artery disease
      - Idiopathic Type 1 diabetes mellitus with peripheral artery disease
    - Idiopathic Type 1 diabetes mellitus with certain specified complications
      - Idiopathic Type 1 diabetes mellitus with diabetic foot
      - Idiopathic Type 1 diabetes mellitus with diabetic skin lesions
      - Idiopathic Type 1 diabetes mellitus with periodontal disease
  - ~~Idiopathic Type 1 diabetes mellitus with unspecified complications~~
  - Idiopathic Type 1 diabetes mellitus without complications

Type 2 diabetes mellitus
- Type 2 diabetes mellitus with coma
  - Type 2 diabetes mellitus with hypoglycaemic coma
  - Type 2 diabetes mellitus with hyperosmolar hyperglycaemic coma without ketoacidosis
- Type 2 diabetes mellitus with acidosis
  - Type 2 diabetes mellitus with ketoacidosis, without coma
  - Type 2 diabetes mellitus with ketoacidosis, with coma
  - Type 2 diabetes mellitus with lactic acidosis
  - Type 2 diabetes mellitus with metabolic acidosis
- Type 2 diabetes mellitus with kidney complications
- Type 2 diabetes mellitus with ophthalmic complications
  - Type 2 diabetes mellitus with retinopathy
    - Type 2 diabetes mellitus with simple retinopathy

Type 2 diabetes mellitus with preproliferative retinopathy
Type 2 diabetes mellitus with proliferative retinopathy
Type 2 diabetes mellitus with macular oedema
Type 2 diabetes mellitus with diabetic cataract
Type 2 diabetes mellitus with glaucoma
Type 2 diabetes mellitus with neurological complications
Type 2 diabetes mellitus with mononeuropathy
Type 2 diabetes mellitus with polyneuropathy
Type 2 diabetes mellitus with other neurological complication
Type 2 diabetes mellitus with macrovascular complications
Type 2 diabetes mellitus with cerebrovascular disease
Type 2 diabetes mellitus with coronary artery disease
Type 2 diabetes mellitus with peripheral artery disease
Type 2 diabetes mellitus with other specified complications
Type 2 diabetes mellitus with diabetic foot
Type 2 diabetes mellitus with diabetic skin lesions
Type 2 diabetes mellitus with periodontal disease
Type 2 diabetes mellitus with unspecified complications
Type 2 diabetes mellitus without complications
Diabetes mellitus, not Type 1 and not Type 2
Diabetes mellitus due to genetic defects of beta cell function
Other specified diabetes mellitus due to genetic defects of beta cell function with coma
Other specified diabetes mellitus due to genetic defects of beta cell function with hypoglycaemic coma
Other specified diabetes mellitus due to genetic defects of beta cell function with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to genetic defects of beta cell function with acidosis
Other specified diabetes mellitus due to genetic defects of beta cell function with ketoacidosis without coma
Other specified diabetes mellitus due to genetic defects of beta cell function with ketoacidosis with coma
Other specified diabetes mellitus due to genetic defects of beta cell function with lactic acidosis
Other specified diabetes mellitus due to genetic defects of beta cell function with metabolic acidosis
Other specified diabetes mellitus due to genetic defects of beta cell function with kidney complications
Other specified diabetes mellitus due to genetic defects of beta cell function with ophthalmic complications
Other specified diabetes mellitus due to genetic defects of beta cell function with retinopathy
Other specified diabetes mellitus due to genetic defects of beta cell function with simple retinopathy
Other specified diabetes mellitus due to genetic defects of beta cell function with preproliferative retinopathy
Other specified diabetes mellitus due to genetic defects of beta cell function with proliferative retinopathy
Other specified diabetes mellitus due to genetic defects of beta cell function with macular oedema
Other specified diabetes mellitus due to genetic defects of beta cell function with diabetic cataract
Other specified diabetes mellitus due to genetic defects of beta cell function with glaucoma
Other specified diabetes mellitus due to genetic defects of beta cell function with neurological complications
Other specified diabetes mellitus due to genetic defects of beta cell function with mononeuropathy
Other specified diabetes mellitus due to genetic defects of beta cell function with polyneuropathy
Other specified diabetes mellitus due to genetic defects of beta cell function with other neurological complication
Other specified diabetes mellitus due to genetic defects of beta cell function with macrovascular complications
Other specified diabetes mellitus due to genetic defects of beta cell function with cerebrovascular disease
Other specified diabetes mellitus due to genetic defects of beta cell function with coronary artery disease
Other specified diabetes mellitus due to genetic defects of beta cell function with peripheral artery disease
Other specified diabetes mellitus due to genetic defects of beta cell function with other specified complications
Other specified diabetes mellitus due to genetic defects of beta cell function with diabetic foot
Other specified diabetes mellitus due to genetic defects of beta cell function with diabetic skin lesions
Other specified diabetes mellitus due to genetic defects of beta cell function with periodontal disease
~~Other specified diabetes mellitus due to genetic defects of beta cell function without complications~~
Other specified diabetes mellitus with unspecified complications
Diabetes mellitus due to genetic defects in insulin action
Other specified diabetes mellitus due to genetic defects in insulin action with coma
Other specified diabetes mellitus due to genetic defects in insulin action with hypoglycaemic coma
Other specified diabetes mellitus due to genetic defects in insulin action with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to genetic defects in insulin action with acidosis
Other specified diabetes mellitus due to genetic defects in insulin action with ketoacidosis without coma
Other specified diabetes mellitus due to genetic defects in insulin action with ketoacidosis with coma
Other specified diabetes mellitus due to genetic defects in insulin action with lactic acidosis
Other specified diabetes mellitus due to genetic defects in insulin action with metabolic acidosis
Other specified diabetes mellitus due to genetic defects in insulin action with kidney complications
Other specified diabetes mellitus due to genetic defects in insulin action with ophthalmic complications
Other specified diabetes mellitus due to genetic defects in insulin action with retinopathy
Other specified diabetes mellitus due to genetic defects in insulin action with simple retinopathy
Other specified diabetes mellitus due to genetic defects in insulin action with preproliferative retinopathy
Other specified diabetes mellitus due to genetic defects in insulin action with proliferative retinopathy
Other specified diabetes mellitus due to genetic defects in insulin action with macular oedema
Other specified diabetes mellitus due to genetic defects in insulin action with diabetic cataract
Other specified diabetes mellitus due to genetic defects in insulin action with glaucoma
Other specified diabetes mellitus due to genetic defects in insulin action with neurological complications
Other specified diabetes mellitus due to genetic defects in insulin action with mononeuropathy
Other specified diabetes mellitus due to genetic defects in insulin action with polyneuropathy
Other specified diabetes mellitus due to genetic defects in insulin action with other neurological complication
Other specified diabetes mellitus due to genetic defects in insulin action with macrovascular complications
Other specified diabetes mellitus due to genetic defects in insulin action with cerebrovascular disease
Other specified diabetes mellitus due to genetic defects in insulin action with coronary artery disease
Other specified diabetes mellitus due to genetic defects in insulin action with peripheral artery disease
Other specified diabetes mellitus due to genetic defects in insulin action with other specified complications
Other specified diabetes mellitus due to genetic defects in insulin action with diabetic foot
Other specified diabetes mellitus due to genetic defects in insulin action with diabetic skin lesions
Other specified diabetes mellitus due to genetic defects in insulin action with periodontal disease
Other specified diabetes mellitus due to genetic defects in insulin action with unspecified complications
Other specified diabetes mellitus due to genetic defects in insulin action without complications
Diabetes mellitus due to diseases of the exocrine pancreas
Other specified diabetes mellitus due to diseases of the exocrine pancreas with coma
Other specified diabetes mellitus due to diseases of the exocrine pancreas with hypoglycaemic coma
Other specified diabetes mellitus due to diseases of the exocrine pancreas with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to diseases of the exocrine pancreas with acidosis
Other specified diabetes mellitus due to diseases of the exocrine pancreas with ketoacidosis without coma
Other specified diabetes mellitus due to diseases of the exocrine pancreas with ketoacidosis with coma
Other specified diabetes mellitus due to diseases of the exocrine pancreas with lactic acidosis
Other specified diabetes mellitus due to diseases of the exocrine pancreas with metabolic acidosis
Other specified diabetes mellitus due to diseases of the exocrine pancreas with kidney complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas with ophthalmic complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas with retinopathy
Other specified diabetes mellitus due to diseases of the exocrine pancreas with simple retinopathy
Other specified diabetes mellitus due to diseases of the exocrine pancreas with preproliferative retinopathy
Other specified diabetes mellitus due to diseases of the exocrine pancreas with proliferative retinopathy
Other specified diabetes mellitus due to diseases of the exocrine pancreas with macular oedema
Other specified diabetes mellitus due to diseases of the exocrine pancreas with diabetic cataract
Other specified diabetes mellitus due to diseases of the exocrine pancreas with glaucoma
Other specified diabetes mellitus due to diseases of the exocrine pancreas with neurological complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas with mononeuropathy

Other specified diabetes mellitus due to diseases of the exocrine pancreas with polyneuropathy
Other specified diabetes mellitus due to diseases of the exocrine pancreas with other neurological complication
Other specified diabetes mellitus due to diseases of the exocrine pancreas with macrovascular complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas with cerebrovascular disease
Other specified diabetes mellitus due to diseases of the exocrine pancreas with coronary artery disease
Other specified diabetes mellitus due to diseases of the exocrine pancreas with peripheral artery disease
Other specified diabetes mellitus due to diseases of the exocrine pancreas with other specified complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas with diabetic foot
Other specified diabetes mellitus due to diseases of the exocrine pancreas with diabetic skin lesions
Other specified diabetes mellitus due to diseases of the exocrine pancreas with periodontal disease
Other specified diabetes mellitus due to diseases of the exocrine pancreas with unspecified complications
Other specified diabetes mellitus due to diseases of the exocrine pancreas without complications
Diabetes mellitus due to endocrinopathies
Other specified diabetes mellitus due to endocrinopathies with coma
Other specified diabetes mellitus due to endocrinopathies with hypoglycaemic coma
Other specified diabetes mellitus due to endocrinopathies with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to endocrinopathies with acidosis
Other specified diabetes mellitus due to endocrinopathies with ketoacidosis without coma
Other specified diabetes mellitus due to endocrinopathies with ketoacidosis with coma
Other specified diabetes mellitus due to endocrinopathies with lactic acidosis
Other specified diabetes mellitus due to endocrinopathies with metabolic acidosis
Other specified diabetes mellitus due to endocrinopathies with kidney complications
Other specified diabetes mellitus due to endocrinopathies with ophthalmic complications
Other specified diabetes mellitus due to endocrinopathies with retinopathy
Other specified diabetes mellitus due to endocrinopathies with simple retinopathy
Other specified diabetes mellitus due to endocrinopathies with preproliferative retinopathy
Other specified diabetes mellitus due to endocrinopathies with proliferative retinopathy
Other specified diabetes mellitus due to endocrinopathies with macular oedema
Other specified diabetes mellitus due to endocrinopathies with diabetic cataract
Other specified diabetes mellitus due to endocrinopathies with glaucoma
Other specified diabetes mellitus due to endocrinopathies with neurological complications
Other specified diabetes mellitus due to endocrinopathies with mononeuropathy
Other specified diabetes mellitus due to endocrinopathies with polyneuropathy
Other specified diabetes mellitus due to endocrinopathies with other neurological complication
Other specified diabetes mellitus due to endocrinopathies with macrovascular complications
Other specified diabetes mellitus due to endocrinopathies with cerebrovascular disease
Other specified diabetes mellitus due to endocrinopathies with coronary artery disease
Other specified diabetes mellitus due to endocrinopathies with peripheral artery disease
Other specified diabetes mellitus due to endocrinopathies with other specified complications
Other specified diabetes mellitus due to endocrinopathies with diabetic foot
Other specified diabetes mellitus due to endocrinopathies with diabetic skin lesions
Other specified diabetes mellitus due to endocrinopathies with periodontal disease
Other specified diabetes mellitus due to endocrinopathies with unspecified complications
Other specified diabetes mellitus due to endocrinopathies without complications
Diabetes mellitus due to drug or chemical
Other specified diabetes mellitus due to drug or chemical with coma
Other specified diabetes mellitus due to drug or chemical with hypoglycaemic coma
Other specified diabetes mellitus due to drug or chemical with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to drug or chemical with acidosis
Other specified diabetes mellitus due to drug or chemical with ketoacidosis without coma
Other specified diabetes mellitus due to drug or chemical with ketoacidosis with coma
Other specified diabetes mellitus due to drug or chemical with lactic acidosis
Other specified diabetes mellitus due to drug or chemical with metabolic acidosis
Other specified diabetes mellitus due to drug or chemical with kidney complications
Other specified diabetes mellitus due to drug or chemical with ophthalmic complications
Other specified diabetes mellitus due to drug or chemical with retinopathy
Other specified diabetes mellitus due to drug or chemical with simple retinopathy
Other specified diabetes mellitus due to drug or chemical with preproliferative retinopathy
Other specified diabetes mellitus due to drug or chemical with proliferative retinopathy
Other specified diabetes mellitus due to drug or chemical with macular oedema
Other specified diabetes mellitus due to drug or chemical with diabetic cataract
Other specified diabetes mellitus due to drug or chemical with glaucoma
Other specified diabetes mellitus due to drug or chemical with neurological complications
Other specified diabetes mellitus due to drug or chemical with mononeuropathy
Other specified diabetes mellitus due to drug or chemical with polyneuropathy
Other specified diabetes mellitus due to drug or chemical with other neurological complication
Other specified diabetes mellitus due to drug or chemical with macrovascular complications
Other specified diabetes mellitus due to drug or chemical with cerebrovascular disease
Other specified diabetes mellitus due to drug or chemical with coronary artery disease
Other specified diabetes mellitus due to drug or chemical with peripheral artery disease
Other specified diabetes mellitus due to drug or chemical with other specified complications
Other specified diabetes mellitus due to drug or chemical with diabetic foot
Other specified diabetes mellitus due to drug or chemical with diabetic skin lesions
Other specified diabetes mellitus due to drug or chemical with periodontal disease
Other specified diabetes mellitus due to drug or chemical with unspecified complications
Other specified diabetes mellitus due to drug or chemical without complications
Diabetes mellitus due to infection
Other specified diabetes mellitus due to infection with coma
Other specified diabetes mellitus due to infection with hypoglycaemic coma
Other specified diabetes mellitus due to infection with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to infection with acidosis
Other specified diabetes mellitus due to infection with ketoacidosis without coma
Other specified diabetes mellitus due to infection with ketoacidosis with coma
Other specified diabetes mellitus due to infection with lactic acidosis
Other specified diabetes mellitus due to infection with metabolic acidosis
Other specified diabetes mellitus due to infection with kidney complications
Other specified diabetes mellitus due to infection with ophthalmic complications
Other specified diabetes mellitus due to infection with retinopathy
Other specified diabetes mellitus due to infection with simple retinopathy
Other specified diabetes mellitus due to infection with preproliferative retinopathy
Other specified diabetes mellitus due to infection with proliferative retinopathy
Other specified diabetes mellitus due to infection with macular oedema
Other specified diabetes mellitus due to infection with diabetic cataract
Other specified diabetes mellitus due to infection with glaucoma
Other specified diabetes mellitus due to infection with neurological complications
Other specified diabetes mellitus due to infection with mononeuropathy
Other specified diabetes mellitus due to infection with polyneuropathy
Other specified diabetes mellitus due to infection with other neurological complication
Other specified diabetes mellitus due to infection with macrovascular complications
Other specified diabetes mellitus due to infection with cerebrovascular disease
Other specified diabetes mellitus due to infection with coronary artery disease
Other specified diabetes mellitus due to infection with peripheral artery disease
Other specified diabetes mellitus due to infection with other specified complications
Other specified diabetes mellitus due to infection with diabetic foot
Other specified diabetes mellitus due to infection with diabetic skin lesions
Other specified diabetes mellitus due to infection with periodontal disease
Other specified diabetes mellitus due to infection with unspecified complications
Other specified diabetes mellitus due to infection without complications
Infections associated with Other specified diabetes mellitus
Diabetes mellitus due to uncommon forms of immune-mediated diabetes
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with coma
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with hypoglycaemic

coma
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with acidosis
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with ketoacidosis without coma
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with ketoacidosis with coma
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with lactic acidosis
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with metabolic acidosis
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with kidney complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with ophthalmic complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with retinopathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with simple retinopathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with preproliferative retinopathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with proliferative retinopathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with macular oedema
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with diabetic cataract
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with glaucoma
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with neurological complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with mononeuropathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with polyneuropathy
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with other neurological complication
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with macrovascular complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with cerebrovascular disease
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with coronary artery disease
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with peripheral artery disease
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with other specified complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with diabetic foot
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with diabetic skin lesions
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with periodontal disease
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes with unspecified complications
Other specified diabetes mellitus due to uncommon forms of immune-mediated diabetes without complications
Other specified diabetes mellitus due to other genetic syndromes
Other specified diabetes mellitus due to other genetic syndromes with coma
Other specified diabetes mellitus due to other genetic syndromes with hypoglycaemic coma
Other specified diabetes mellitus due to other genetic syndromes with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to other genetic syndromes with acidosis
Other specified diabetes mellitus due to other genetic syndromes with ketoacidosis without coma
Other specified diabetes mellitus due to other genetic syndromes with ketoacidosis with coma
Other specified diabetes mellitus due to other genetic syndromes with lactic acidosis
Other specified diabetes mellitus due to other genetic syndromes with metabolic acidosis
Other specified diabetes mellitus due to other genetic syndromes with kidney complications
Other specified diabetes mellitus due to other genetic syndromes with ophthalmic complications
Other specified diabetes mellitus due to other genetic syndromes with retinopathy
Other specified diabetes mellitus due to other genetic syndromes with simple retinopathy
Other specified diabetes mellitus due to other genetic syndromes with preproliferative retinopathy
Other specified diabetes mellitus due to other genetic syndromes with proliferative retinopathy
Other specified diabetes mellitus due to other genetic syndromes with macular oedema
Other specified diabetes mellitus due to other genetic syndromes with diabetic cataract
Other specified diabetes mellitus due to other genetic syndromes with glaucoma
Other specified diabetes mellitus due to other genetic syndromes with neurological complications
Other specified diabetes mellitus due to other genetic syndromes with mononeuropathy
Other specified diabetes mellitus due to other genetic syndromes with polyneuropathy
Other specified diabetes mellitus due to other genetic syndromes with other neurological complication
Other specified diabetes mellitus due to other genetic syndromes with macrovascular complications
Other specified diabetes mellitus due to other genetic syndromes with cerebrovascular disease
Other specified diabetes mellitus due to other genetic syndromes with coronary artery disease
Other specified diabetes mellitus due to other genetic syndromes with peripheral artery disease
Other specified diabetes mellitus due to other genetic syndromes with other specified complications
Other specified diabetes mellitus due to other genetic syndromes with diabetic foot
Other specified diabetes mellitus due to other genetic syndromes with diabetic skin lesions
Other specified diabetes mellitus due to other genetic syndromes with periodontal disease
Other specified diabetes mellitus due to other genetic syndromes with unspecified complications
Other specified diabetes mellitus due to other genetic syndromes without complications
Diabetes mellitus due to clinically defined subtypes/syndromes
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with coma
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with hypoglycaemic coma
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with hyperosmolar hyperglycaemic coma without ketoacidosis
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with acidosis
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with ketoacidosis without coma
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with ketoacidosis with coma
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with lactic acidosis
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with metabolic acidosis
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with kidney complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with ophthalmic complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with retinopathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with simple retinopathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with preproliferative retinopathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with proliferative retinopathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with macular oedema
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with diabetic cataract
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with glaucoma
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with neurological complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with mononeuropathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with polyneuropathy
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with other neurological complication
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with macrovascular complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with cerebrovascular disease
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with coronary artery disease
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with peripheral artery disease
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with other specified complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with diabetic foot
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with diabetic skin lesions
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with periodontal disease
Other specified diabetes mellitus due to clinically defined subtypes/syndromes with unspecified complications
Other specified diabetes mellitus due to clinically defined subtypes/syndromes without complications
Diabetes mellitus without mention of type
Diabetes mellitus without mention of type with coma
Unspecified diabetes mellitus with hypoglycaemic coma

- Unspecified diabetes mellitus hyperosmolar hyperglycaemic coma without ketoacidosis
- Diabetes mellitus without mention of type with acidosis
  - Unspecified diabetes mellitus with ketoacidosis without coma
  - Unspecified diabetes mellitus with lactic acidosis
  - Unspecified diabetes mellitus with metabolic acidosis
  - Unspecified diabetes mellitus with ketoacidosis with coma
- Diabetes mellitus without mention of type with kidney complications
- *Unspecified diabetes mellitus due to genetic defects of beta cell function with ophthalmic complications*
  - Unspecified diabetes mellitus with retinopathy
    - Unspecified diabetes mellitus with simple retinopthy
    - Unspecified diabetes mellitus with preproliferative retinopathy
    - Unspecified diabetes mellitus with proliferative retinopathy
  - Unspecified diabetes mellitus with macular oedema
  - Unspecified diabetes mellitus with diabetic cataract
  - Unspecified diabetes mellitus with glaucoma
- Diabetes mellitus without mention of type with neurological complications
  - Unspecified diabetes mellitus with mononeuropathy
  - Unspecified diabetes mellitus with polyneuropathy
  - ~~Unspecified diabetes mellitus with other neurological complication~~
- Diabetes mellitus without mention of type with macrovascular complications
  - Unspecified diabetes mellitus with cerebrovascular disease
  - Unspecified diabetes mellitus with coronary artery disease
  - Unspecified diabetes mellitus with peripheral artery disease
- Diabetes mellitus without mention of type with certain specified complications
  - Unspecified diabetes mellitus with diabetic foot
  - Unspecified diabetes mellitus with diabetic skin lesions
  - Unspecified diabetes mellitus with periodontal disease
- Diabetes mellitus without mention of type with unspecified complications
- Diabetes mellitus without mention of type without complications

Impaired glucose regulation
- ~~Impaired glucose tolerance with unspecified complication~~
- Impaired fasting glucose (IFG)
- Impaired glucose tolerance (IGT)
  - Impaired glucose tolerance with macrovascular complications
    - Impaired glucose tolerance with cerebrovascular complications
    - Impaired glucose tolerance with coronary artery disease
    - Impaired glucose tolerance with peripheral artery disease
- Impaired glucose tolerance with multiple complications
- Impaired glucose tolerance without complication

Insulin-resistance syndromes
- Insulin-resistance syndrome type A
- Insulin-resistance syndrome type B
- Rabson-Mendenhall syndrome
- Laminopathy type Decaudain-Vigouroux
- Leprechaunism
- HAIR-AN syndrome
- Insulin resistance - short fifth metacarpals
- Berardinelli-Seip congenital lipodystrophy
- Familial partial lipodystrophy
  - Familial partial lipodystrophy, Köbberling type
  - Familial partial lipodystrophy, Dunnigan type
  - Familial partial lipodystrophy associated with PPARG mutations
  - Familial partial lipodystrophy due to AKT2 mutations
  - Familial partial lipodystrophy due to lamin-A mutations
  - Familial partial lipodystrophy associated with PLIN1 mutations
- Acquired generalized lipodystrophy

Rare specified diabetes mellitus
- Maternally inherited diabetes and deafness (MIDD)
- *Thiamine-responsive megaloblastic anaemia syndrome*
- Woodhouse-Sakati syndrome
- Mitochondrial myopathy with diabetes mellitus
- Wolfram syndrome
- *MODY syndrome*
  - MODY 1 syndrome
  - MODY 2 syndrome
  - MODY 3 syndrome
  - MODY 4 syndrome
  - MODY 5 syndrome
  - MODY 6 syndrome
  - MODY 7 syndrome
  - MODY 8 syndrome
- Wolcott-Rallison syndrome

Diabetes mellitus in pregnancy
- Diabetes mellitus in pregnancy: Pre-existing diabetes mellitus, insulin-dependent
- Diabetes mellitus in pregnancy: Pre-existing diabetes mellitus, non-insulin-dependent
- Diabetes mellitus in pregnancy: Pre-existing malnutrition-related diabetes mellitus
- ~~Diabetes mellitus in pregnancy: Pre-existing diabetes mellitus, unspecified~~
- Diabetes mellitus arising in pregnancy
- ~~Diabetes mellitus in pregnancy, unspecified~~

Neonatal diabetes mellitus
- *Transient neonatal diabetes mellitus*
- Permanent neonatal diabetes mellitus
- Syndromic permanent neonatal diabetes mellitus
  - DEND syndrome
  - Permanent neonatal diabetes mellitus - pancreatic and cerebellar agenesis

Other disorders of glucose regulation and pancreatic internal secretion

Nondiabetic hypoglycaemic coma

Certain specified disorders of pancreatic internal secretion
- Nondiabetic hypoglycaemia
- Other hypoglycaemia
  - Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)
    - *Diffuse hyperinsulinism*
      - Diffuse hyperinsulinism, diazoxide-resistant
      - Diffuse hyperinsulinism, diazoxide-sensitive
    - Exercise-induced hyperinsulinism
    - *Hyperinsulinism due to focal adenomatous hyperplasia*
  - Transient hyperinsulinemic hypoglycemia of newborn
  - Insulinoma
  - Hypoglycaemia
- ~~Hypoglycaemia, unspecified~~
- Increased secretion of glucagon
  - Hyperplasia of pancreatic endocrine cells with glucagon excess
  - *Glucagonoma*
- Abnormal secretion of gastrin
  - Drug-induced hypergastrinaemia
  - Zollinger-Ellison syndrome
    - Anastomotic ulcer due to Zollinger-Ellison syndrome
    - Gastric ulcer due to Zollinger-Ellison syndrome
    - Duodenal ulcer due to Zollinger-Ellison syndrome
  - Gastrinoma
    - Zollinger-Ellison syndrome
      - Anastomotic ulcer due to Zollinger-Ellison syndrome
      - Gastric ulcer due to Zollinger-Ellison syndrome
      - *Duodenal ulcer due to Zollinger-Ellison syndrome*

- ~~Other specified disorders of pancreatic internal secretion~~
  - Increased secretion from endocrine pancreas of: growth hormone-releasing hormone
  - Increased secretion from endocrine pancreas of: pancreatic polypeptide
  - Increased secretion from endocrine pancreas of: somatostatin
  - Increased secretion from endocrine pancreas of: vasoactive-intestinal polypeptide
  - Pancreatic islet cells; hyperplasia, alpha cells
- ~~Disorder of pancreatic internal secretion, unspecified~~

Disorders of the parathyroids and parathyroid hormone system

- Hypoparathyroidism
  - Cataract in hypoparathyroidism
    - Cataract in idiopathic hypoparathyroidism
    - Cataract in pseudohypoparathyroidism
  - Hypoparathyroidism due to impaired PTH secretion
  - Idiopathic hypoparathyroidism
    - Cataract in idiopathic hypoparathyroidism
  - Pseudohypoparathyroidism
    - Albright hereditary osteodystrophy
      - Pseudohypoparathyroidism type 1A
    - Cataract in pseudohypoparathyroidism
    - Pseudohypoparathyroidism type 1B
    - Pseudohypoparathyroidism type 1C
    - Pseudohypoparathyroidism type 2
    - Pseudopseudohypoparathyroidism
  - Primary hypoparathyroidism
    - Familial isolated hypoparathyroidism
      - Autosomal dominant hypocalcemia
      - Familial isolated hypoparathyroidism due to agenesis of parathyroid gland
        - X-linked recessive hypoparathyroidism
        - Hypoparathyroidism by glial cells missing B [GCMB] mutations
      - Familial isolated hypoparathyroidism due to impaired parathormone secretion
    - Hypoparathyroidism due to destruction of the parathyroid glands
      - Hypoparathyroidism after iodine thyroid ablation
      - Hypoparathyroidism due to external radiation
      - Hypoparathyroidism due to surgical removal of parathyroid gland
      - Hypoparathyroidism associated with haemochromatosis
      - Hypoparathyroidism associated with Wilson's disease
    - Autoimmune hypoparathyroidism
  - Transient neonatal hypoparathyroidism due to maternal hyperparathyroidism
  - Secondary hypoparathyroidism
    - Hypoparathyroidism associated with granulomatous disease
  - Syndrome with hypoparathyroidism
    - Hypoparathyroidism - deafness - renal disease
    - Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
    - Pearson syndrome
    - CATCH 22 phenotype [22q11.2 deletion]
    - Kenny-Caffey syndrome, autosomal dominant
    - Kenny-Caffey syndrome, autosomal recessive - Sanjad-Sakati syndrome
    - Kearns-Sayre syndrome
  - ~~Other hypoparathyroidism~~
    - familial hypoparathyroidism
  - ~~Hypoparathyroidism, unspecified~~
- Hyperparathyroidism
  - Primary hyperparathyroidism
    - Familial primary hyperparathyroidism
      - Familial isolated hyperparathyroidism
      - Familial parathyroid adenoma
      - Hyperparathyroidism - jaw tumour syndrome
      - Primary parathyroid hyperplasia
    - Neonatal severe primary hyperparathyroidism
    - Non-familial primary hyperparathyroidism
      - Hyperplasia of parathyroid
      - ~~Malignant neoplasm: Parathyroid gland~~
      - Benign neoplasm of parathyroid gland
  - Secondary hyperparathyroidism
  - Other hyperparathyroidism
  - Hyperparathyroidism, unspecified
  - Familial hypocalciuric hypercalcaemia
  - Acquired hypocalciuric hypercalcemia
  - Tertiary hyperparathyroidism
- Certain specified disorders of parathyroid gland
  - Polyneuropathy in other specified disorders of parathyroid gland
  - cyst of parathyroid gland
  - hemorrhage of parathyroid gland
- ~~Disorder of parathyroid gland, unspecified~~

Disorders of the pituitary hormone system

- Hyperfunction of pituitary gland
  - ~~Syndrome of inappropriate antidiuretic hormone secretion~~
  - Acromegaly and pituitary gigantism
    - Cardiomyopathy associated with acromegaly
    - Somatotroph adenoma
    - Arthropathy associated with acromegaly
  - Hyperprolactinaemia
    - Prolactinoma
    - Drug-induced hyperprolactinaemia
    - Hypothyroidism-associated hyperprolactinemia
    - Functional hyperprolactinoma
      - Chiari-Frommel Syndrome
      - Argonz-del Castillo Syndrome
    - Hyperprolactinemia caused by other hypothalamo-pituitary disorders
    - Macroprolactinemia
  - Syndrome of inappropriate secretion of antidiuretic hormone
    - Pituitary inappropriate secretion of antidiuretic hormone
    - Ectopic inappropriate secretion of antidiuretic hormone
  - Central precocious puberty
  - ~~Other hyperfunction of pituitary gland~~
- Hypofunction and certain specified disorders of pituitary gland
  - Hypopituitarism
    - Non-acquired hypopituitarism
      - Isolated FSH deficiency
      - Non-acquired combined hypopituitarism
        - Panhypopituitarism
        - Septo-optic dysplasia
          - Optic nerve hypoplasia associated with endocrinologic deficiencies
      - Nonacquired isolated growth hormone deficiency
        - Growth hormone deficiency with or without pituitary stalk interruption syndrome (PSIS)
          - Genetic growth hormone deficiency
          - Idiopathic growth hormone deficiency
        - Short stature due to growth hormone qualitative anomaly
      - Pituitary stalk interruption syndrome
      - Short stature due to a defect in growth hormone receptor or post-receptor pathway
        - Growth delay due to insulin-like growth factor I deficiency
        - Growth delay due to insulin-like growth factor I resistance
        - Short stature due to growth hormone resistance
        - Short stature due to primary acid-labile subunit deficiency

- Laron syndrome with immunodeficiency
- Congenital hypogonadotropic hypogonadism
  - Anosmic congenital hypogonadotropic hypogonadism
  - Normosmic congenital hypogonadotropic hypogonadism
  - Syndrome with hypogonadotropic hypogonadism
    - Bardet-Biedl syndrome
    - CHARGE syndrome
    - Cataract - intellectual deficit - hypogonadism
    - Prader-Willi syndrome
    - Woodhouse-Sakati syndrome
  - Hypogonadotropic hypogonadism associated with other endocrinopathies
    - Obesity due to congenital leptin deficiency
    - Obesity due to leptin receptor gene deficiency
    - Obesity due to prohormone convertase-1 deficiency
- Acquired hypopituitarism
  - Drug-induced hypopituitarism
  - Tumoral hypopituitarism
  - Infectious hypopituitarism
  - Autoimmune hypopituitarism
  - Vascular hypopituitarism
  - Sheehan syndrome
  - Hypopituitarism secondary to a metabolic disease
  - *Hypopituitarism secondary to a granulomatous disease*
  - Traumatic hypopituitarism
  - Hypophysitis
    - Lymphocytic hypophysitis
    - Granulomatous hypophysitis
    - IgG4-related hypophysitis
    - Xanthomatous hypophysitis
  - Pineal germinoma
  - Suprasellar germinoma
  - Chordoma
  - Pituitary deficiency due to Rathke pouch cysts
  - Pituitary dermoid and epidermoid cysts
  - Pituitary deficiency due to empty sella turcica syndrome
  - Pituitary apoplexy
  - Late-onset isolated ACTH deficiency
  - Idiopathic hypopituitarism
  - Isolated prolactin deficiency
- Central diabetes insipidus
  - Wolfram syndrome
  - Acquired central diabetes insipidus
    - Acquired central diabetes due to Infiltrative disease
    - Idiopathic acquired central diabetes insipidus
    - Traumatic central diabetes insipidus
  - *Non-acquired central diabetes insipidus*
- Hypothalamic dysfunction, not elsewhere classified
  - Diencephalic syndrome
- Pituitary incidentaloma
- ~~Other disorders of pituitary gland~~
  - Adiposogenital dystrophy
  - Non-secreting pituitary adenoma
  - abscess of pituitary
  - cerebralis; adiposis
  - *colloid cyst of pituitary gland*
  - cyst; craniobuccal pouch
  - cyst; craniopharyngeal pouch
  - cyst; hypophysis
  - cyst; intrasellar
  - hypophysis infarct
  - hypothalamic infantilism with obesity syndrome
  - launois-cleret syndrome
  - pituitary embolism
  - pituitary gland cyst
  - pituitary gland degeneration
  - pituitary gland hemorrhage
  - pituitary gland hypertrophy
  - pituitary gland infarction
  - pituitary obesity
  - rathke cleft cyst
- ~~Disorder of pituitary gland, unspecified~~

Disorders of the adrenal glands and adrenal hormone system

- Cushing syndrome
  - ACTH-dependent Cushing syndrome
    - Pituitary-dependent Cushing disease
    - Ectopic ACTH syndrome
  - ACTH-independent Cushing syndrome
    - *ACTH-independent Cushing syndrome due to bilateral adrenocortical hyperplasia*
      - ACTH-independent macronodular adrenal hyperplasia
      - Primary pigmented nodular adrenocortical disease
      - Carney complex
    - ACTH-independent Cushing syndrome due to unilateral tumour
      - Adrenocortical adenoma
      - Adrenocortical carcinoma
    - Drug-induced Cushing syndrome
  - Pseudo-Cushing syndrome
    - *Pseudo-Cushing syndrome of other causes*
    - Alcohol-induced pseudo-Cushing syndrome
  - Nelson syndrome
  - ~~Other Cushing syndrome~~
- Adrenogenital disorders
  - 46,XX disorders of sex development induced by androgens of fetal origin
    - Glucocorticoid resistance
    - Congenital adrenal hyperplasia
      - Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency
      - Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency
      - Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
        - Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, classic form
          - Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, classic form, salt wasting
          - Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, classic form, simple virilizing
        - Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, nonclassic form
      - Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase deficiency
      - Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency
      - Congenital adrenal hyperplasia due to side-chain cleavage enzyme deficiency
      - Congenital lipoid adrenal hyperplasia due to STAR deficiency
        - Congenital lipoid adrenal hyperplasia due to STAR deficiency, classic form
        - Congenital lipoid adrenal hyperplasia due to STAR deficiency, nonclassic form
  - *46,XX disorders of sex development induced by androgens of maternal origin*
    - 46,XX androgen-induced disorder of sex development due to maternal adrenal hyperplasia
    - 46,XX androgen-induced disorder of sex development due to maternal androluteoma
    - 46,XX androgen-induced disorders of sex development due to a virilizing maternal ovarian or adrenal tumour
      - 46,XX androgen-induced disorder of sex development due to maternal Krukenberg tumor

- 46,XX androgen-induced disorder of sex development due to maternal adrenal tumor
- 46,XX androgen-induced disorder of sex development due to maternal arrhenoblastoma
- 46,XX androgen-induced disorders of sex development of iatrogenic maternal origin
- 46,XX disorder of sex development due to synthetic oral progestagen or diethylstilbestrol
- 46,XX disorder of sex development due to testosterone or related steroids
- Certain specified adrenogenital disorders
- Drug-induced adrenogenital disorders
- Hyperandrogenism due to cortisone reductase deficiency
- Premature adrenarche
- ~~Adrenogenital disorder, unspecified~~
- Hyperaldosteronism
- Primary hyperaldosteronism
- Familial hyperaldosteronism
- Familial hyperaldosteronism type 1
- Familial hyperaldosteronism type 2
- Familial hyperaldosteronism type 3
- Non-familial primary hyperaldosteronism
- Aldosterone-producing adenoma
- Aldosterone-producing carcinoma
- Idiopathic hyperaldosteronism
- Primary bilateral adrenal hyperplasia
- Primary unilateral adrenal hyperplasia
- Secondary hyperaldosteronism
- ~~Other hyperaldosteronism~~
- aldosteronism with hyperplasia of the adrenal cortex
- bartter's syndrome
- juxtaglomerular hyperplasia with secondary aldosteronism
- Hypoaldosteronism
- Familial hypoaldosteronism
- Familial hyperreninaemic hypoaldosteronism type 1
- Familial hyperreninaemic hypoaldosteronism type 2
- Adrenocortical insufficiency
- Congenital adrenocortical insufficiency
- Congenital adrenal hypoplasia
- Autosomal recessive congenital adrenal hypoplasia
- X-linked congenital adrenal hypoplasia
- Congenital isolated ACTH deficiency
- Cytomegalic congenital adrenal hypoplasia
- Familial adrenal hypoplasia
- Familial glucocorticoid deficiency
- Neonatal adrenoleukodystrophy
- IMAGe syndrome
- Triple A syndrome
- Achalasia - alacrimia syndrome
- X-linked adrenoleukodystrophy
- X-linked cerebral adrenoleukodystrophy
- Adrenomyeloneuropathy
- Acquired adrenocortical insufficiency
- Adrenal crisis
- Adrenal haemorrhage
- Adrenal infarction
- Interruption of corticosteroid therapy
- Waterhouse-Friderichsen syndrome
- Drug-induced adrenocortical insufficiency
- Adrenomedullary hyperfunction
- Adrenomedullary hyperplasia
- Secreting paraganglioma
- Von Hippel-Lindau disease
- Phaeochromocytoma
- Adrenal incidentaloma
- Certain specified disorders of adrenal gland
- ~~Disorder of adrenal gland, unspecified~~
- Apparent mineralocorticoid excess

Disorders of the gonadal hormone system

- Ovarian dysfunction
- Premature ovarian failure
- Primary amenorrhoea
- 46,XX gonadal dysgenesis
- Secondary amenorrhoea
- Premature ovarian failure due to autoimmune oophoritis
- ~~Other ovarian dysfunction~~
- hyperhormonal; amenorrhea
- ovarian hyperfunction
- ovarian hyperthecosis
- ovarian; hypergonadism
- hyperovarism
- Clinical Hyperandrogenism
- Drug-induced androgen excess
- Tumoral androgen excess
- Hirsutism
- Polycystic ovarian syndrome
- HAIR-AN syndrome
- Polycystic ovary
- Anovulation
- Anovulation or oligo-ovulation
- Diminished ovarian reserve
- Ovarian Hyper Stimulation Syndrome (OHSS)
- Testicular dysfunction and testosterone-related disorders
- Testicular hyperfunction
- Testotoxicosis
- Testicular hypofunction
- Male infertility due to a systemic disease
- Male infertility with normal virilization due to acquired testicular defects
- Defective biosynthesis of testosterone in aged men
- Male infertility associated with autoimmunity
- Male infertility associated with drug
- Male infertility associated with environmental toxin
- Male infertility associated with mycoplasma infection
- Male infertility associated with radiation
- Male infertility associated with spinal cord injury
- Male infertility with normal virilization due to developmental or structural testicular defects
- Chromosome Y deletion
- Male infertility associated with cryptorchidism
- Male infertility associated with varicocele
- Sertoli cell only syndrome
- 46,XY disorders of sex development
- 46,XY disorders of sex development due to a defect in testicular development
- Dysgenetic 46,XY disorder of sex development
- WAGR syndrome [11p13 deletion]
- Alpha thalassaemia - X-linked intellectual deficit
- Campomelic dysplasia
- Denys-Drash syndrome
- Frasier syndrome
- 46,XY gonadal dysgenesis

- Complete 46,XY gonadal dysgenesis
- Partial 46,XY gonadal dysgenesis
- 46,XY gonadal dysgenesis - motor and sensory neuropathy
- Embryonic testicular regression syndrome
- Sudden infant death - dysgenesis of the testes
- Testicular agenesis
- 46,XY disorder of sex development due to a defect in testosterone metabolism
- 46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
- 46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency
- 46,XY disorder of sex development due to isolated 17, 20 lyase deficiency
- Leydig cell hypoplasia
- LH resistance due to complete LH receptor inactivation
- LH resistance due to partial LH receptor inactivation
- Smith-Lemli-Opitz syndrome
- 46,XY disorder of sex development due to androgen resistance
- Complete peripheral androgen resistance syndrome
- Partial peripheral androgen resistance syndrome
- Persistent Müllerian duct syndrome
- Fuqua-Berkovitz syndrome
- Congenital non genetic 46,XY disorder of sex development
- 46,XY disorder of sex development due to environnemental chemical exposure
- 46,XY disorder of sex development due to maternal ingestion of progestagen and estrogen

Disorders of puberty, not elsewhere classified
- Aromatase deficiency
- Disorder of puberty; oestrogen resistance
- Delayed puberty
- Peripheral precocious puberty
- Testotoxicosis
- Aromatase excess syndrome
- McCune-Albright syndrome
- Certain specified disorders of puberty
- Premature thelarche
- precocious thelarche
- ~~Disorder of puberty, unspecified~~

Polyglandular dysfunction
- Multiple polyglandular tumours
- Carney complex
- Carney triad
- Carney-Stratakis syndrome
- Multiple endocrine neoplasia type 1
- Multiple endocrine neoplasia type 2
- Multiple endocrine neoplasia type 2A
- Multiple endocrine neoplasia type 2B
- Multiple endocrine neoplasia type 4
- Von Hippel-Lindau disease
- Autoimmune polyendocrinopathy
- Autoimmune polyendocrinopathy type 1
- Autoimmune polyendocrinopathy type 2
- Autoimmune polyendocrinopathy type 3
- Autoimmune polyendocrinopathy type 4
- Immune dysregulation - polyendocrinopathy - enteropathy, X-linked
- Polyglandular hyperfunction
- ~~Certain specified polyglandular dysfunction~~
- dystrophy; polyglandular
- pluriglandular; atrophy
- polyglandular; deficiency
- polyglandular; sclerosis
- ~~Polyglandular dysfunction, unspecified~~

Certain endocrine disorders
- Endocrine tumours
- Bronchial endocrine tumour
- Cutaneous neuroendocrine carcinoma
- Gastrointestinal endocrine tumours
- Gastric endocrine tumour
- Neuroendocrine neoplasm of duodenum
- Neuroendocrine tumour of duodenum
- Neurodendocrine carcinoma of duodenum
- Other specified neuroendocrine neoplasm of duodenum
- Neuroendocrine tumour of small intestine
- Carcinoid and other neuroendocrine tumour of jejunum
- Carcinoid and other neuroendocrine tumour of ileum
- Carcinoid and other neuroendocrine tumour of Meckel's diverticulum
- Carcinoid and other neuroendocrine tumour of overlapping lesion of small intestine
- Neuroendocrine carcinoma of small intestine
- Neuroendocrine carcinoma of jejunum
- Neuroendocrine carcinoma of ileum
- Neuroendocrine carcinoma of Meckel's diverticulum
- Neuroendocrine carcinoma of overlapping lesion of small intestine
- Colon neuroendocrine neoplasm
- Neuroendocrine neoplasms of appendix
- Neuroendocrine tumour of appendix
- Neuroendocrine carcinoma of appendix
- Other specified neurodendocrine neoplasms of appendix
- Rectal neuroendocrine neoplasm
- Anal endocrine tumour
- Laryngeal endocrine tumour
- Middle ear endocrine tumour
- Thymic endocrine tumour
- Neuroendocrine neoplasms of pancreas
- Neuroendocrine tumours of pancreas
- Neuroendocrine carcinoma of pancreas
- Insulino
- ma
- Glucagonoma
- Gastrinoma
- Zollinger-Ellison syndrome
- Anastomotic ulcer due to Zollinger-Ellison syndrome
- Gastric ulcer due to Zollinger-Ellison syndrome
- Duodenal ulcer due to Zollinger-Ellison syndrome
- VIPoma
- Somatostatinoma
- Other neuroendocrine tumour of pancreas
- GRFoma
- PPoma
- Hepatic endocrine tumour
- Gallbladder neuroendocrine tumour
- ~~Carcinoid syndrome~~
- Carcinoid syndrome
- Certain specified hypersecretion of intestinal hormones
- Ectopic hormone secretion, not elsewhere classified
- Short stature, not elsewhere classified
- Short stature due to growth hormone resistance
- Constitutional tall stature
- ~~Certain specified endocrine disorders~~

- ~~Endocrine disorder, unspecified~~
- Disorders of lipoprotein metabolism and certain specified lipidaemias
  - Hyperlipoproteinaemia
    - Hypercholesterolaemia
      - Primary hypercholesterolaemia
        - Familial hypercholesterolaemia
          - Familial hypercholesterolaemia - heterozygous
          - Familial hypercholesterolaemia - homozygous
        - Familial defective apolipoprotein B-100
        - Autosomal recessive hypercholesterolaemia
        - Autosomal dominant hypercholesterolaemia
        - Polygenic hypercholesterolaemia
        - Sitosterolaemia
      - Secondary hypercholesterolaemia
    - Hyperglyceridaemia
      - Major hypertriglyceridaemia
        - Hyperlipoproteinaemia type 1
          - Familial apolipoprotein C-II deficiency
          - Familial lipoprotein lipase deficiency
        - Hyperlipoproteinaemia type 4
          - Familial hyperlipoproteinaemia type 4
          - Secondary hyperliporpoteinaemia type 4
        - Hyperlipoproteinaemia type 5
          - Familial hyperlipoproteinaemia type 5
          - Secondary hyperlipoproteinaemia type 5
    - Mixed hyperlipidaemia
      - Hyperlipidaemia type 3
        - Hyperlipidaemia type 3 - Apolipoprotein E deficiency
        - Hyperlipidaemia type 3 - familial defective apolipoprotein B
        - Hyperlipidaemia type 3 - familial dysbetalipoproteinemia
        - Hyperlipidaemia type 3 - familial dysilpoproteinaemia
        - Secondary hyperlipoproteinaemia type 3
      - Mixed hyperlipidaemia - type IIb hyperlipoproteinaemia
        - Familial combined hyperlipidaemia
        - Secondary combined hyperlipidaemia
      - Mixed hyperlipidaemia due to hepatic triglyceride lipase deficiency
    - Hyperalphalipoproteinaemia
      - Familial hyperalphalipoproteinaemia
      - Hyperalphalipoproteinaemia due to cholesteryl ester transfer protein deficiency
      - Hyperlipidaemia due to cubilin deficiency
    - ~~Other hyperlipidaemia~~
      - Familial combined hyperlipidaemia
    - ~~Hyperlipidaemia, unspecified~~
  - Hypolipoproteinaemia
    - Hypoalphalipoproteinaemia
      - Familial hypoalphalipoproteinaemia
        - Apolipoprotein A-I deficiency
        - Familial lecithin-cholesterol acyltransferase deficiency
          - Fish-eye disease
        - Analphalipoproteinaemia
      - Secondary hypoalphalipoproteinaemia
    - Hypobetalipoproteinaemia
      - Abetalipoproteinaemia
      - Familial hypobetalipoproteinaemia
        - Benign familial hypobetalipoproteinaemia
        - Truncated form of apolipoprotein B-100
      - Chylomicron retention disease
      - Secondary hypobetalipoproteinaemia
    - Lecithin-cholesterol acyltransferase deficiency
      - Norum disease
  - ~~Other disorders of lipoprotein metabolism~~
    - lipoid proteinosis
    - multicentric reticulohistiocytosis
    - nicolau-balus syndrome
    - lipogranulomatosis
  - ~~Disorder of lipoprotein metabolism, unspecified~~
  - Lipoid dermatoarthritis
- Nutritional diseases
- Metabolic disorders
  - Inborn errors of metabolism
    - Disorders of amino acid and other organic acid metabolism
      - Disorders of phenylalanine metabolism
        - Phenylketonuria
          - Classical phenylketonuria
            - Classical phenylketonuria, phenylalanine hydroxylase partial deficency
            - Classical phenylketonuria, phenylalanine hydroxylase total deficiency
          - Nonclassical phenylketonuria
          - Mild hyperphenylalaninaemia
          - Phenylketonuria, BH4 deficiency
        - Maternal phenylketonuria
      - Disorders of tyrosine metabolism
        - Alkaptonuria
          - Endogenous ochronosis
        - Tyrosinaemia type 1
        - Tyrosinaemia type 2
        - Tyrosinaemia type 3
        - Hawkinsinuria
        - Transient tyrosine oxidase deficiency
        - Autosomal recessive dopa-responsive dystonia
          - Tyrosine hydroxylase deficiency
        - Albinism
          - Ocular albinism
            - X-linked recessive ocular albinism
            - Ocular albinism - late-onset sensorineural deafness
          - Oculocutaneous albinism
            - Oculocutaneous albinism type 1A
            - Oculocutaneous albinism type 1B
              - Yellow oculocutaneous albinism
              - Minimal pigment oculocutaneous albinism
              - Platinum oculocutaneous albinism
              - Temperature-sensitive oculocutaneous albinism
            - Oculocutaneous albinism type 2
              - Tyrosinase-positive oculocutaneous albinism
              - Brown oculocutaneous albinism
            - Oculocutaneous albinism type 3
            - Oculocutaneous albinism type 4
            - Hermansky-Pudlak syndrome
              - Hermansky-Pudlak syndrome type 7
              - Hermansky-Pudlak syndrome type 8
              - Hermansky-Pudlak syndrome type 9
              - Hermansky-Pudlak syndrome with neutropenia
              - Hermansky-Pudlak syndrome with pulmonary fibrosis
              - Hermansky-Pudlak syndrome without pulmonary fibrosis
            - Chédiak-Higashi syndrome

- Oculocerebral syndrome with hypopigmentation
- Autosomal recessive dopa-responsive dystonia
  - Tyrosine hydroxylase deficiency
- Disorders of histidine metabolism
  - Histidinaemia
  - Urocanic aciduria
  - Formiminoglutamic aciduria
- Disorders of tryptophan metabolism
  - Hypertryptophanaemia
  - Encephalopathy due to hydroxykynureninuria
  - Pellagra-like skin rash - neurological manifestations
- Disorders of lysine and hydroxylysine metabolism
  - Hyperlysinaemia
    - Hyperlysinaemia type 1
    - Hyperlysinaemia type 2
  - 2-aminoadipic aciduria
  - 2-ketoadipic aciduria
  - Seizures - intellectual deficit due to hydroxylysinuria
- Disorders of the gamma-glutamyl cycle
  - Oxoprolinuria
  - Gamma-glutamyl transpeptidase deficiency
  - Cysteinylglycinase deficiency
  - Haemolytic anaemia due to glutathione synthetase deficiency
    - Glutathione synthetase deficiency with 5-oxoprolinuria
    - Glutathione synthetase deficiency without 5-oxoprolinuria
  - Haemolytic anaemia due to gamma-glutamylcysteine synthetase deficiency
- Disorders of serine metabolism
  - 3-phosphoglycerate dehydrogenase deficiency
  - 3-phosphoserine phosphatase deficiency
  - Phosphoserine aminotransferase deficiency
- Disorders of glycine metabolism
  - Glycine encephalopathy
    - Neonatal glycine encephalopathy
    - Infantile glycine encephalopathy
    - Atypical glycine encephalopathy
  - Sarcosinaemia
  - D-glyceric acidaemia
- Disorders of proline and hydroxyproline metabolism
  - Hyperhydroxyprolinaemia
  - Hyperprolinaemia type 1
  - Hyperprolinaemia type 2
  - Progressive neurodegeneration - joint laxity - cataract
- Disorders of ornithine metabolism
  - Hyperornithinaemia
  - Hyperornithinaemia-hyperammonaemia-homocitrullinuria
- Disorders of urea cycle metabolism
  - Argininosuccinic aciduria
  - Carbamoylphosphate synthetase deficiency
  - Argininaemia
  - Citrullinaemia
    - Citrullinaemia type 1
      - Acute neonatal citrullinaemia type 1
      - Adult-onset citrullinaemia type 1
    - Citrin deficiency
      - Citrullinaemia type 2
      - Neonatal intrahepatic cholestasis caused by citrin deficiency
  - Ornithine carbamoyltransferase deficiency
  - Hyperammonaemia due to N-acetylglutamate synthetase deficiency
  - Transient hyperammonaemia of the newborn
  - Hyperornithinaemia-hyperammonaemia-homocitrullinuria
  - Hyperinsulinism-hyperammonaemia syndrome
  - Other disorders of the urea cycle
  - Unspecified hyperammonaemia
- Disorders of methionine cycle and sulfur amino acid metabolism
  - Brain demyelination due to methionine adenosyltransferase deficiency
  - Glycine N-methyltransferase deficiency
  - Psychomotor retardation and myopathy due to S-adenosylhomocysteine hydrolase deficiency
  - Cystathioninuria
  - Classical homocystinuria
  - Homocystinuria without methylmalonic aciduria
    - Methylcobalamin deficiency type cbl E
    - Methylcobalamin deficiency type cbl G
    - Methylcobalamin deficiency type cbl Dv1
  - Encephalopathy due to sulfite oxidase deficiency
    - Isolated sulfite oxidase deficiency
    - Sulfite oxidase deficiency due to molybdenum cofactor deficiency
      - Molybdenum cofactor deficiency, complementation group A
      - Molybdenum cofactor deficiency, complementation group B
      - Molybdenum cofactor deficiency, complementation group C
  - Congenital megaloblastic anaemia due to transcobalamin II deficiency
  - Hypermethioninaemia due to adenosine kinase deficiency
  - Other genetic defects of methionine cycle and sulfur amino acid metabolism
  - Secondary non-genetic disorders of methionine cycle and sulfur amino acid metabolism
- Disorders of beta and omega amino acid metabolism
  - Gamma aminobutyric acid transaminase deficiency
- Disorders of branched-chain amino acid metabolism
  - Leucinosis
  - Methioninaemia
  - Maple-syrup-urine disease
    - Classical maple-syrup-urine disease
    - Intermediate maple-syrup-urine disease
    - Intermittent maple-syrup-urine disease
    - Thiamine-responsive maple-syrup-urine disease
  - Hyperleucine-isoleucinaemia
  - Hypervalinaemia
  - Acetoacetyl-CoA thiolase deficiency
    - Ketoacidosis due to beta-ketothiolase deficiency
    - Mitochondrial 2-methylacetoacetyl-Coa thiolase deficiency, non potassium-stimulated
    - Cytosolic acetoacetyl-CoA thiolase deficiency
  - Certain specified disorders of branched-chain amino acid metabolism
- Organic acidurias
  - Classical organic acidurias
    - Ketoacidosis due to beta-ketothiolase deficiency
    - Methylmalonic aciduria - homocystinuria
      - Methylmalonic aciduria - homocystinuria type cbl C
      - Methylmalonic aciduria - homocystinuria type cbl D
      - Methylmalonic aciduria - homocystinuria type cbl F
    - Methylmalonic aciduria
      - Methylmalonic aciduria, vitamin B12 unresponsive
        - Complete deficiency of methylmalonyl-CoA mutase
        - Partial deficiency of methylmalonyl-CoA mutase
        - Methylmalonyl-CoA epimerase deficiency
        - Methylmalonyl-CoA epimerase deficiency with sepiapterin reductase deficiency
      - Methylmalonic aciduria, vitamin B12 responsive

- Vitamin B12 responsive methylmalonic aciduria type cbl A
- Vitamin B12 responsive methylmalonic aciduria type cbl B
- Vitamin B12 responsive methylmalonic aciduria type cbl mut-
- Propionic aciduria
- Isovaleric aciduria
- Multiple carboxylase deficiency
  - Multiple carboxylase deficiency due to biotinidase deficiency
  - Multiple carboxylase deficiency due to holocarboxylase synthetase deficiency
- Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency
- 3-hydroxyisobutyric aciduria
- 3-hydroxy-3-methylglutaric aciduria
- 3-methylcrotonylglycinuria
- 3-methylglutaconic aciduria
  - 3-methylglutaconic aciduria type 1
  - Barth syndrome
  - 3-methylglutaconic aciduria type 3
  - 3-methylglutaconic aciduria type 4
  - 3-methylglutaconic aciduria type 5
- 2-methylbutyric aciduria
- Isobutyric aciduria
- Methylmalonate semialdehyde dehydrogenase deficiency
- Glutaric aciduria type 3

Cerebral organic acidurias
- Glutaric aciduria type 1
- 2-hydroxyglutaric aciduria
  - L-2-hydroxyglutaric aciduria
  - D-2-hydroxyglutaric aciduria
    - D-2-hydroxyglutarate dehydrogenase deficiency
    - Mitochondrial isocitrate dehydrogenase deficiency
- Aminoacylase deficiency
  - Neurological conditions associated with aminoacylase 1 deficiency
  - Canavan disease
- Malonic aciduria
- Ethylmalonic encephalopathy
- 4-hydroxybutyric aciduria

Disorders of peptide metabolism
- Prolidase deficiency
- Carnosinaemia
- Homocarnosinosis

Certain specificied disorders of amino acid and other organic acid metabolism
- Congenital brain dysgenesis due to glutamine synthetase deficiency
- Trimethylaminuria

Disorders of carbohydrate metabolism

Disorders of the pentose phosphate pathway
- Essential pentosuria
- Ribose-5-phosphate isomerase deficiency
- Transaldolase deficiency
- Anaemia due to glucose-6-phosphate dehydrogenase deficiency

Disorders of glycerol metabolism
- Glycerol kinase deficiency
  - Isolated glycerol kinase deficiency
  - Glycerol kinase deficiency - contiguous gene syndrome
- Glycerol intolerance

Disorders of glyoxylate metabolism
- Primary hyperoxaluria type 1
- Primary hyperoxaluria type 2
- Hyperoxaluria non1-non2 type

Other specified disorders of carbohydrate metabolism
- Triose phosphate-isomerase deficiency

Glycogen storage diseases
- Glycogen storage disease due to glycogen synthase deficiency
  - Glycogen storage disease due to liver glycogen synthase deficiency
  - Glycogen storage disease due to muscle and heart glycogen synthase deficiency
- Glycogen storage disease due to glucose-6-phosphate system deficiency
  - Glycogen storage disease due to glucose-6-phosphate transport defect
  - Glycogen storage disease due to glucose-6-phosphatase deficiency
- Glycogen storage disease due to acid maltase deficiency
  - Glycogen storage disease due to acid maltase deficiency, infantile onset
  - Glycogen storage disease due to acid maltase deficiency, juvenile onset
  - Glycogen storage disease due to acid maltase deficiency, adult onset
- Glycogen storage disease due to LAMP-2 deficiency
- Glycogen storage disease due to glycogen debranching enzyme deficiency
- Glycogen storage disease due to glycogen branching enzyme deficiency
- Glycogen storage disease due to muscle glycogen phosphorylase deficiency
- Glycogen storage disease due to liver glycogen phosphorylase deficiency
- Glycogen storage disease due to muscle phosphofructokinase deficiency
- Glycogen storage disease due to glycogen phosphorylase kinase deficiency
  - Glycogen storage disease due to liver glycogen phosphorylase kinase deficiency
  - Glycogen storage disease due to liver and muscle glycogen phosphorylase kinase deficiency
  - Glycogen storage disease due to heart glycogen phosphorylase kinase deficiency
  - Glycogen storage disease due to muscle glycogen phosphorylase kinase deficiency
- Glycogen storage disease due to muscle phosphoglycerate mutase deficiency
- Glycogen storage disease due to GLUT2 deficiency
- Glycogen storage disease due to aldolase A deficiency
- Glycogen storage disease due to muscle beta-enolase deficiency
- Glycogen storage disease due to phosphoglucomutase deficiency
- Glycogen storage disease due to glycogenin deficiency
- Glycogen storage disease due to phosphoglycerate kinase 1 deficiency
- Glycogen storage disease due to lactate dehydrogenase deficiency
- Glycogen storage disease due to muscle pyruvate kinase deficiency
- Unspecified glycogen storage disease

Disorders of galactose metabolism
- Galactosaemia
  - Galactose-1-phosphate uridyltransferase deficiency
  - Uridine diphosphate galactose-4-epimerase deficiency
    - Erythrocyte uridine diphosphate galactose-4-epimerase deficiency
    - Generalised uridine diphosphate galactose-4-epimerase deficiency
  - Galactokinase deficiency

Disorders of fructose metabolism
- Essential fructosuria
- Fructose-1,6-diphosphatase deficiency
- Hereditary fructose intolerance

Disorders of lipid metabolism

Disorders of fatty acid oxidation and ketone body metabolism
- Adrenoleukodystrophy
  - Adult-onset autosomal dominant leukodystrophy
  - Zellweger syndrome
  - Neonatal adrenoleukodystrophy
  - X-linked adrenoleukodystrophy
    - X-linked cerebral adrenoleukodystrophy
    - Adrenomyeloneuropathy
- Disorders of carnitine transport and the carnitine cycle
  - Carnitine transporter deficiency
  - Carnitine palmitoyltransferase I deficiency

- Carnitine palmitoyltransferase II deficiency
  - Carnitine palmitoyltransferase II deficiency, myopathic form
  - Carnitine palmitoyltransferase II deficiency, severe infantile form
  - Carnitine palmitoyltransferase II deficiency, neonatal form
- Carnitine-acylcarnitine translocase deficiency

Disorders of mitochondrial fatty acid oxidation
- Acyl-CoA dehydrogenase deficiencies
  - Short chain acyl-CoA dehydrogenase deficiency
  - Medium chain acyl-CoA dehydrogenase deficiency
  - Long chain acyl-CoA dehydrogenase deficiency
  - Very long chain acyl-CoA dehydrogenase deficiency
  - Multiple acyl-CoA dehydrogenase deficiency
    - Electron transfer flavoprotein deficiency, alpha chain
    - Electron transfer flavoprotein deficiency, beta chain
    - Electron transfer flavoprotein-ubiquinone oxidoreductase deficiency
- 3-hydroxyacyl-CoA dehydrogenase deficiencies
  - Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency
  - Medium and short chain 3-hydroxyacyl-CoA dehydrogenase deficiency
  - Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- Mitochondrial trifunctional protein deficiency

Disorders of ketone body metabolism
- Cytosolic acetoacetyl-CoA thiolase deficiency
- 3-hydroxy 3-methylglutaryl-CoA lyase deficiency
- Succinyl-CoA acetoacetate transferase deficiency

Other disorders of fatty acid oxidation and ketone body metabolism
- Malonic aciduria
- Sjögren-Larsson syndrome

Disorders of sterol metabolism

Disorders of cholesterol synthesis
- Mevalonate kinase deficiency
  - Mevalonic aciduria
  - Mevalonate kinase [MVK] deficiency with recurrent fever
- Smith-Lemli-Opitz syndrome
- Desmosterolosis
- Lathosterolosis
- Cholesterol-ester transfer protein deficiency
- Chondrodysplasia punctata, X-linked dominant
- Greenberg dysplasia
- Congenital hemidysplasia with ichthyosiform erythroderma and limbs defects [CHILD]

Other disorders of sterol metabolism
- Sitosterolaemia
- Lethal neonatal chondrodysplasia punctata
- X-linked ichthyosis

Bile acid synthesis defect with cholestasis
- Cerebrotendinous xanthomatosis
- Congenital bile acid synthesis defect type 1
- Congenital bile acid synthesis defect type 2
- Congenital bile acid synthesis defect type 3
- Congenital bile acid synthesis defect type 4

Neutral lipid storage disease
- Dorfman-Chanarin disease
- Neutral lipid storage myopathy

Disorders of energy metabolism

Disorders of pyruvate metabolism

Pyruvate kinase deficiency
- Glycogen storage disease due to muscle pyruvate kinase deficiency
- Haemolytic anaemia due to red cell pyruvate kinase deficiency

Phosphoenolpyruvate carboxykinase deficiency
- Phosphoenolpyruvate carboxykinase 1 deficiency
- Phosphoenolpyruvate carboxykinase 2 deficiency

Lactate dehydrogenase deficiency

Pyruvate dehydrogenase complex deficiency
- Pyruvate dehydrogenase complex E1-alpha subunit deficiency
- Pyruvate dehydrogenase complex E1-beta subunit deficiency
- Pyruvate dehydrogenase complex E2 subunit deficiency
- Pyruvate dehydrogenase complex E3 subunit deficiency
- Pyruvate dehydrogenase complex E3-binding protein deficiency
- Pyruvate dehydrogenase phosphatase deficiency

Pyruvate carboxylase deficiency

Disorders of the citric acid cycle

Fumarase deficiency

Alpha-ketoglutarate dehydrogenase deficiency

Aconitase deficiency

Disorders of mitochondrial oxidative phosphorylation

Mitochondrial oxidative phosphorylation disorders due to mitochondrial DNA anomalies
- Proximal tubulopathy - diabetes mellitus - cerebellar ataxia
- Recessive mitochondrial ataxic syndrome
- Large-scale single deletions of mitochondrial DNA
  - Pearson syndrome
  - Chronic progressive external ophthalmoplegia
  - Kearns-Sayre syndrome
- Point mutations of mitochondrial DNA
  - Maternally inherited diabetes and deafness (MIDD)
  - Mitochondrial encephalopathy with lactic acidosis and stroke-like episodes (MELAS)
  - Neuropathy, ataxia, and retinitis pigmentosa
  - Myoclonic epilepsy - ragged red fibres (MERRF)
  - Leber hereditary optic neuropathy (LHON)
    - Leber 'plus' disease
  - Maternally inherited Leigh syndrome (MILS)
  - Sporadic Leigh syndrome
  - Maternally inherited mitochondrial dystonia
  - Maternally inherited mitochondrial cardiomyopathy
  - Maternally inherited mitochondrial myopathy
    - 'Pure' mitochondrial myopathy
    - Lethal infantile mitochondrial myopathy
    - Mitochondrial myopathy with diabetes mellitus
    - Mitochondrial myopathy with reversible cytochrome C oxidase deficiency
  - Non-syndromic mitochondrial sensorineural deafness
- Duplications of mitochondrial DNA

Mitochondrial oxidative phosphorylation disorders due to nuclear DNA anomalies
- Mitochondrial DNA depletion syndromes
  - Mitochondrial DNA depletion syndrome, hepatocerebral form
    - Infantile onset spinocerebellar ataxia
    - Navajo neurohepatopathy
  - Mitochondrial DNA depletion syndrome, myopathic form
  - Mitochondrial DNA depletion syndrome, encephalomyopathic form
    - Encephalomyopathic mitochondrial DNA depletion syndrome with methylmalonic aciduria
    - Encephalomyopathic mitochondrial DNA depletion syndrome with renal tubulopathy
    - Mitochondrial encephalomyopathy - aminoacidopathy
  - Myoneurogastrointestinal encephalopathy syndrome (MNGIE)
  - Fatal infantile lactic acidosis with methylmalonic aciduria
  - Childhood-onset autosomal dominant optic atrophy

- Multiple mitochondrial DNA deletion syndromes
  - Progressive external ophthalmoplegia, autosomal dominant
    - Progressive external ophthalmoplegia, autosomal dominant, type 1
    - Progressive external ophthalmoplegia, autosomal dominant, type 2
    - Progressive external ophthalmoplegia, autosomal dominant, type 3
    - Progressive external ophthalmoplegia, autosomal dominant, type 4
    - Progressive external ophthalmoplegia, autosomal dominant, type 5
  - Progressive external ophthalmoplegia, autosomal recessive
  - Mitochondrial DNA deletion ataxia neuropathy spectrum
    - Mitochondrial recessive ataxic syndrome (MIRAS)
    - Sensory ataxic neuropathy - dysarthria - ophthalmoparesis (SANDO)
    - Spinocerebellar ataxia with epilepsy (SCAE)
  - Autosomal dominant optic atrophy plus syndrome
  - Deafness - optic atrophy syndrome
  - Autosomal dominant optic atrophy and cataract
- Coenzyme Q10 deficiency
  - Early-onset ataxia with oculomotor apraxia and hypoalbuninaemia
  - Deafness - encephaloneuropathy - obesity - valvulopathy
  - Cerebellar atrophy - ataxia - seizures
- GRACILE syndrome
- Björnstad syndrome
- Cardioencephalopathy with hyperammonaemia
- Exercise intolerance with lactic acidosis
- Isolated oxidative phosphorylation defects with variable phenotype, not elsewhere classified
  - Mitochondrial respiratory chain complex I structural subunit gene defect
  - Mitochondrial respiratory chain complex I assembly gene defect
  - Mitochondrial respiratory chain complex II structural subunit gene defect
  - Mitochondrial respiratory chain complex II assembly gene defect
  - Mitochondrial respiratory chain complex III structural subunit gene defect
  - Mitochondrial respiratory chain complex III assembly gene defect
  - Mitochondrial respiratory chain complex IV structural subunit gene defect
  - Mitochondrial respiratory chain complex IV assembly gene defect
  - ATP synthase structural subunit gene defect
  - ATP synthase assembly gene defect
- Mitochondrial protein translation defects
  - Pontocerebellar hypoplasia type 6
  - Combined oxidative phosphorylation deficiency
    - Combined oxidative phosphorylation deficiency type 1
    - Combined oxidative phosphorylation deficiency type 2
    - Combined oxidative phosphorylation deficiency type 3
    - Combined oxidative phosphorylation deficiency type 4
    - Combined oxidative phosphorylation deficiency type 5
    - Combined oxidative phosphorylation deficiency type 6
    - Combined oxidative phosphorylation deficiency type 7
  - Mitochondrial myopathy with sideroblastic anaemia (MLASA)
    - Mitochondrial myopathy with sideroblastic anaemia, type 1 (MLASA1)
    - Mitochondrial myopathy with sideroblastic anaemia, type 2 (MLASA2)
  - Acute infantile liver failure due to mitochondrial DNA-encoded proteins synthesis defect
  - Leukoencephalopathy with brain stem - spinal cord involvement - lactate elevation
- Renal tubulopathy - encephalopathy - liver failure
- Leigh syndrome
  - Infantile subacute necrotizing encephalopathy
  - Leigh syndrome due to cytochrome C oxidase deficiency
  - Leigh syndrome with cardiomyopathy
  - Leigh syndrome with leukodystrophy
  - Leigh syndrome with nephrotic syndrome
  - Saguenay-Lac-Saint-Jean cytochrome C oxidase deficiency
  - Dystonia due to Leigh syndrome
  - Maternally inherited Leigh syndrome (MILS)
  - Sporadic Leigh syndrome
- Mitochondrial oxidative phosphorylation disorders with no known genetic basis
  - Autosomal recessive optic atrophy type 7
  - Deafness - opticoacoustic nerve atrophy - dementia
  - FASTKD2-related infantile mitochondrial encephalomyopathy
  - Succinic acidaemia
  - Zellweger-like syndrome without peroxisomal anomalies
  - Isolated mitochondrial respiratory chain complex I deficiency
  - Isolated mitochondrial respiratory chain complex II deficiency
  - Isolated mitochondrial respiratory chain complex III deficiency
  - Isolated mitochondrial respiratory chain complex IV deficiency
    - Ataxia - leukodystrophy - tubulopathy, due to cytochrome C oxidase deficiency
    - Pancreatic insufficiency - anaemia - hyperostosis
  - Isolated ATP synthase deficiency
- Disorders of mitochondrial membrane transport
  - Mitochondrial substrate carrier disorders
    - Cardiomyopathy - hypotonia - lactic acidosis
    - Mitochondrial phosphate carrier deficiency
    - Mitochondrial aspartate glutamate carrier 1 deficiency
    - Mitochondrial glutamate carrier 1 deficiency
    - Autosomal recessive sideroblastic anaemia, pyridoxine-refractory
  - Mitochondrial protein import disorders
    - Mohr-Tranebjærg syndrome
    - Deafness-dystonia optic atrophy syndrome
- Unspecified mitochondrial disorders
  - Ethylmalonic encephalopathy
  - Leigh syndrome with no known genetic or respiratory chain deficiency
  - X-linked sideroblastic anaemia - ataxia
  - Cataract - cardiomyopathy
- Disorders of creatine metabolism
  - X-linked creatine transporter deficiency
  - Guanidinoacetate methyltransferase deficiency
  - Arginine:glycine amidinotransferase deficiency
- Other disorders of energy metabolism
  - Glucose phosphate isomerase deficiency
    - Haemolytic anaemia due to glucose-phosphate isomerase deficiency
  - Lactate transporter deficiency
  - Lipoamide dehydrogenase deficiency
  - Haemolytic anaemia due to red cell diphosphoglycerate mutase deficiency
- Congenital disorders of glycosylation and certain specified disorders of protein modification
  - Disorders of protein N-glycosylation
    - Phosphomannomutase 2 deficiency
    - Phosphomannose isomerase deficiency
    - Glucosyltransferase 1 deficiency
    - Mannosyltransferase 6 deficiency
    - Mannosyltransferase 8 deficiency
    - Glucosyltransferase 2 deficiency
    - Mannosyltransferase 2 deficiency
    - Dolichyl-phosphate N-acetylgalactosamine phosphotransferase deficiency
    - Mannosyltransferase 1 deficiency
    - Mannosyltransferase 7-9 deficiency
    - M5-DLO flippase deficiency
    - N-acetylglucosaminyltransferase deficiency
    - Glucosidase 1 deficiency
    - TUSC3 congenital disorder of glycosylation

- SRD5A3 congenital disorder of glycosylation
- DPM3 congenital disorder of glycosylation
- ALG11 congenital disorder of glycosylation
- *Disorders of protein O-glycosylation*
  - Defects in O-xylosylglycan synthesis
    - Beta-1,4-galactosyltransferase 7 deficiency
    - *Multiple congenital exostoses*
  - Defects in O-N-acetylgalactosaminylglycan synthesis
    - Polypeptide N-acetylgalactosaminyl transferase deficiency
    - *Hyperphosphataemic familial tumoral calcinosis*
  - Defects in O-xylosyl/N-acetylgalactosaminylglycan synthesis
    - Schneckenbecken dysplasia
  - *Defects in O-mannosylglycan synthesis*
    - Protein-O-mannosyltransferase 1
    - Protein-O-mannosyltransferase 2 deficiency
    - *Protein-O-mannose beta-1,2-N-acetyglucosaminyltransferase deficiency*
    - Fukutin deficiency
    - Fukutin-related protein deficiency
    - N-acetylglucosaminyl*transferase-like protein deficiency*
    - O-fucose-specific beta-1,3-N-acetylglucosaminyltransferase deficiency
    - O-fucose-specific beta-1,3-N-glucosyltransferase deficiency
- Disorders of glycosphingolipid and glycosylphosphatidylinositol anchor glycosylation
  - Lactosylceramide alpha-2,3-sialyltransferase deficiency
  - Phosphatidylinositolglycan, class M deficiency
- Disorders of multiple glycosylation and other pathways
  - Dolichol-phosphate-mannose synthase 1 deficiency
  - Mannose-P-dolichol utilisation defect 1
  - Beta-1,4-galactosyltransferase deficiency
  - Hereditary inclusion body myositis
  - CMP-sialic acid transporter deficiency
  - Leukocyte adhesion deficiency type 2
  - Dolichol kinase deficiency
  - Conserved oligomeric Golgi complex deficiencies
    - Component of conserved oligomeric Golgi complex 1 deficiency
    - Component of conserved oligomeric Golgi complex 4 deficiency
    - Component of conserved oligomeric Golgi complex 5 deficiency
    - Component of conserved oligomeric Golgi complex 7 deficiency
    - Component of conserved oligomeric Golgi complex 8 deficiency
  - V-ATPase deficiencies
- Disorders of protein ubiquitinylation
- Other specified disorders of protein modification

Disorders of purine, pyrimidine and nucleotide metabolism

- Disorders of purine metabolism
  - AICAR transformylase deficiency
  - Adenine phosphoribosyltransferase deficiency
    - Adenine phosphoribosyltransferase deficiency type I
    - Adenine phosphoribosyltransferase deficiency type II
  - Adenosine monophosphate deaminase deficiency
  - Adenylosuccinate lyase deficiency
  - Deoxyguanosine kinase deficiency
  - Familial juvenile hyperuricaemic nephropathy
  - Hypoxanthine-guanine phosphoribosyltranferase deficiency
    - Kelley-Seegmiller syndrome
    - Lesch-Nyhan syndrome
  - Inosine triphosphate pyrophospholydrolase deficiency
  - Myoadenylate deaminase deficiency
  - Phosphoribosyl pyrophosphate synthase 1 defects
    - Lethal ataxia with deafness and optic atrophy
    - Phosphoribosyl pyrophosphate synthase superactivity
    - X-linked Charcot-Marie-Tooth disease type 5
  - Thiopurine S-methyltransferase deficiency
  - Xanthinuria
    - Xanthinuria type 1
    - Xanthinuria type 2
  - Primary gout
    - Acute primary gout
      - Acute primary gout, monoarticular
      - Acute primary gout, polyarticular
    - Chronic primary gout
      - Chronic primary gout, monoarticular
      - Chronic primary gout, polyarticular
  - Immunodeficiency due to purine nucleoside phosphorylase deficiency
  - Severe combined immunodeficiency [SCID] T- B- due to adenosine deaminase deficiency
  - *Haemolytic anaemia due to adenosine deaminase excess*
- Disorders of pyrimidine metabolism
  - Beta-aminoisobutyrate-pyruvate transaminase deficiency
  - Beta-ureidopropionase deficiency
  - Dihydroorotate dehydrogenase deficiency
  - Dihydropyrimidinase deficiency
  - Dihydropyrimidine dehydrogenase deficiency
  - Hereditary orotic aciduria
    - Hereditary orotic aciduria type 1
    - Hereditary orotic aciduria type 2
  - Thymidine kinase 2 deficiency
  - Thymidine phosphorylase deficiency
  - Uridine-5'-monophosphate hydrolase superactivity
  - Hyper-beta-alaninaemia
  - Haemolytic anaemia due to pyrimidine 5' nucleotidase deficiency
- Disorders of nucleotide metabolism
  - Aicardi-Goutières syndrome
    - Aicardi-Goutières syndrome type 1
    - Aicardi-Goutières syndrome type 2
    - Aicardi-Goutières syndrome type 3
    - Aicardi-Goutières syndrome type 4
    - Aicardi-Goutières syndrome type 5
  - Cystic leukoencephalopathy without megalencephaly
  - Haemolytic anaemia due to adenosine triphosphatase deficiency
- Hyperuricaemia without signs of inflammatory arthritis and tophaceous disease
- ~~Other disorders of purine and pyrimidine metabolism~~
- ~~Disorder of purine and pyrimidine metabolism, unspecified~~

Lysosomal diseases

- Sphingolipidosis
  - Gangliosidosis
    - GM1 gangliosidosis
      - GM1 gangliosidosis type 1
      - GM1 gangliosidosis type 2
      - GM1 gangliosidosis type 3
    - GM2 gangliosidosis
      - Retinal dystrophy in GM2 gangliosidosis
      - Sandhoff disease
        - Infantile Sandhoff disease
        - Juvenile Sandhoff disease
        - Adult Sandhoff disease
      - Tay-Sachs disease

- B variant of hexosaminidase A deficiency
- B1 variant of hexosaminidase A deficiency
- Hexosaminidase activator deficiency
- Mucolipidosis type 4
- Gaucher disease
  - Gaucher disease type 1
  - Gaucher disease type 2
  - Gaucher disease type 3
  - Gaucher disease - ophthalmoplegia - cardiovascular calcification
  - Perinatal-lethal Gaucher disease
  - Atypical Gaucher disease due to saposin C deficiency
- Krabbe disease
  - Infantile Krabbe disease
  - Late-infantile or juvenile Krabbe disease
  - Adult Krabbe disease
- Fabry disease
  - Fabry disease; X-linked
  - Fabry disease; autosomal dominant
  - Fabry disease; autosomal recessive
  - Angiokeratoma corporis diffusum
  - Glomerular disease associated with Fabry disease
- Metachromatic leukodystrophy
  - Arylsulfatase A deficiency
    - Late infantile metachromatic leukodystrophy
    - Juvenile metachromatic leukodystrophy
    - Adult metachromatic leukodystrophy
  - Metachromatic leukodystrophy due to sphingolipid activator protein I deficiency
- Mucosulfatidosis
- Niemann-Pick disease
  - Niemann-Pick disease type A
  - Niemann-Pick disease type B
  - Niemann-Pick disease type C
    - Niemann-Pick disease type C, severe perinatal
    - Niemann-Pick disease type C, severe early infantile neurologic onset
    - Niemann-Pick disease type C, late infantile neurologic onset
    - Niemann-Pick disease type C, juvenile neurologic onset
    - Niemann-Pick disease type C, adult neurologic onset
- Farber disease
- Lysosomal acid lipase deficiency
  - ~~Wolman disease~~
  - Cholesteryl ester storage disease
  - Wolman disease
- Encephalopathy due to prosaposin deficiency

- Neuronal ceroid lipofuscinosis
  - Congenital neuronal ceroid lipofuscinosis
  - Infantile neuronal ceroid lipofuscinosis
  - Late infantile neuronal ceroid lipofuscinosis
  - Juvenile neuronal ceroid lipofuscinosis
  - Adult neuronal ceroid lipofuscinosis
- Pycnodysostosis
- Glycoproteinosis
  - Mucolipidosis
    - Mucolipidosis type 3
    - Mucolipidosis type 4
    - Mucolipidosis type 2
  - Oligosaccharidosis
    - Aspartylglucosaminuria
    - Alpha-mannosidosis
      - Infantile alpha-mannosidosis
      - Adult alpha-mannosidosis
    - Beta-mannosidosis
    - Fucosidosis
    - Sialidosis
      - Sialidosis type 1
      - Sialidosis type 2
        - Congenital sialidosis type 2
        - Juvenile sialidosis type 2
    - Alpha-N-acetylgalactosaminidase deficiency
      - Alpha-N-acetylgalactosaminidase deficiency type 1
      - Alpha-N-acetylgalactosaminidase deficiency type 2
      - Alpha-N-acetylgalactosaminidase deficiency type 3
    - Galactosialidosis
- Mucopolysaccharidosis
  - Mucopolysaccharidosis type 1
    - Mucopolysaccharidosis type 1H
    - Mucopolysaccharidosis type 1H/S
    - Mucopolysaccharidosis type 1S
  - Mucopolysaccharidosis type 2
    - Mucopolysaccharidosis type 2A
    - Mucopolysaccharidosis type 2B
  - Mucopolysaccharidosis type 3
    - Mucopolysaccharidosis type 3A
    - Mucopolysaccharidosis type 3B
    - Mucopolysaccharidosis type 3C
    - Mucopolysaccharidosis type 3D
  - Mucopolysaccharidosis type 4
    - Mucopolysaccharidosis type 4A
    - Mucopolysaccharidosis type 4B
  - Mucopolysaccharidosis type 6
  - Mucopolysaccharidosis type 7
  - Mucopolysaccharidosis type 9
- Disorders of sialic acid metabolism
  - Free sialic acid storage disease
  - Sialuria

- Peroxisomal diseases
  - Liver disease due to peroxisomal diseases
  - Disorders of peroxisome biogenesis
    - Zellweger syndrome
    - Zellweger spectrum disorder, mild form
      - Neonatal adrenoleukodystrophy
      - Infantile Refsum disease
    - Zellweger spectrum disorder, unclassified clinical severity
      - PEX1 deficiency
      - PEX2 deficiency
      - PEX3 deficiency
      - PEX5 deficiency
      - PEX6 deficiency
      - PEX10 deficiency
      - PEX12 deficiency
      - PEX13 deficiency
      - PEX14 deficiency
      - PEX16 deficiency
      - PEX19 deficiency
      - PEX26 deficiency

- Disorders of peroxisomal alpha-, beta- and omega-oxidation
  - Refsum disease
    - Infantile Refsum disease
  - Congenital bile acid synthesis defect type 4
  - X-linked adrenoleukodystrophy
    - X-linked cerebral adrenoleukodystrophy
    - Adrenomyeloneuropathy
  - Peroxisomal D-bifunctional enzyme deficiency
  - Peroxisomal acyl-CoA oxidase deficiency
  - Sterol carrier protein deficiency
- Other peroxisomal disorders
  - Pseudo-Zellweger syndrome
  - Primary hyperoxaluria type 1
  - Glutaric aciduria type 3
  - Acatalasaemia
- Rhizomelic chondrodysplasia punctata
  - Rhizomelic chondrodysplasia punctata type 1
  - Rhizomelic chondrodysplasia punctata type 2
  - Rhizomelic chondrodysplasia punctata type 3
- Disorders of porphyrin and haem metabolism
- Disorders of bilirubin metabolism and excretion
  - Arthrogryposis - renal dysfunction - cholestasis
  - Crigler-Najjar syndrome
    - Crigler-Najjar syndrome type 1
    - Crigler-Najjar syndrome type 2
  - Gilbert syndrome
  - Dubin-Johnson syndrome
  - Rotor syndrome
  - Progressive familial intrahepatic cholestasis
    - Progressive familial intrahepatic cholestasis type 1
    - Progressive familial intrahepatic cholestasis type 2
    - Progressive familial intrahepatic cholestasis type 3
  - Benign recurrent intrahepatic cholestasis
    - Benign intrahepatic cholestasis type 1
    - Benign intrahepatic cholestasis type 2
- Porphyrias
  - Non-acute porphyrias
    - Porphyria cutanea tarda
      - Sporadic porphyria cutanea tarda
      - Familial porphyria cutanea tarda
      - Toxic porphyria cutanea tarda
    - Hepatoerythropoietic porphyria
    - Erythropoietic protoporphyria
      - Erythropoietic protoporphyria due to ferrochelatase deficiency
      - Erythropoietic protoporphyria due to gain of function of erythroid-specific aminolevulinic acid synthase 2
    - Congenital erythropoietic porphyria
  - Acute porphyrias
    - Acute intermittent porphyria
    - Homozygous acute intermittent porphyria
    - Variegate porphyria
    - Homozygous variegate porphyria
    - Hereditary coproporphyria
    - Homozygous hereditary coproporphyria
    - Harderoporphyria
    - ALAD porphyria
- X-linked sideroblastic anaemia, pyridoxine-responsive
- Disorders of neurotransmitter metabolism
- Disorders of biogenic amine metabolism
  - Disorders of catecholamine synthesis
    - Aromatic L-amino acid decarboxylase deficiency
    - Dopamine beta-hydroxylase deficiency
  - Disorders of pterin metabolism
    - Malignant hyperphenylalaninaemia due to tetrahydrobiopterin deficiency
      - Dihydropteridine reductase deficiency
      - 6-pyruvoyl-tetrahydropterin synthase deficiency
      - GTP cyclohydrolase 1 deficiency
      - Pterin-4-carbinolamine dehydratase deficiency
      - Motor and cognitive disorder due to sepiapterin reductase deficiency
    - Dopa-responsive dystonia
      - Autosomal recessive dopa-responsive dystonia
        - Tyrosine hydroxylase deficiency
      - Autosomal dominant dopa-responsive dystonia
      - Autosomal recessive dopa-responsive dystonia
        - Tyrosine hydroxylase deficiency
  - Monoamine oxidase-A deficiency
- Disorders of gamma aminobutyric acid metabolism
  - Gamma aminobutyric acid transaminase deficiency
  - 4-hydroxybutyric aciduria
- Disorders of pyridoxine metabolism
  - Pyridoxine dependent epilepsy with antiquitin mutations
  - Pyridoxal dependent epilepsy
- Metabolic disease involving other neurotransmitter deficiency
  - Folinic acid-responsive seizures
  - Hereditary hyperekplexia
- Butyrylcholinesterase deficiency
- Certain specified inborn errors of metabolism
- Autosomal dominant myoglobinuria
- Genetic recurrent myoglobinuria
- Alpha-1-antitrypsin deficiency
  - Alpha-1 antitrypsin deficiency panniculitis
- Disorders of metabolite absorption and transport
- Disorders of amino acid absorption and transport
- Hyperdibasic aminoaciduria type 2
- Hypotonia-cystinuria syndrome
- Oculocerebrorenal syndrome
  - Glaucoma in Lowe's syndrome
- Hyperdibasic aminoaciduria type 1
- Lysinuric protein intolerance
- Neutral amino acid transport defect
  - Hartnup syndrome
  - Iminoglycinuria
- Cystinosis
  - Renal tubulo-interstitial disorders in cystinosis
  - Juvenile nephropatic cystinosis
  - Benign adult nephropathic cystinosis
- Cystinuria
  - Cystinuria type A
  - Cystinuria type B
- Blue diaper syndrome
- Dicarboxylic aminoaciduria
- Disorders of carbohydrate absorption and transport
- Glucose-galactose malabsorption
- Maltase-glucoamylase deficiency

- Congenital sucrase-isomaltase deficiency
- Alpha, alpha trehalase deficiency
- Fructose malabsorption
- Acquired monosaccharide malabsorption
- Disorders of facilitated glucose transport
  - Glycogen storage disease due to GLUT2 deficiency
  - Encephalopathy due to GLUT1 deficiency
  - Familial renal glucosuria
- Other disorders of intestinal carbohydrate absorption
- Lactose intolerance
  - Primary lactase deficiency
  - Congenital lactase deficiency
  - Secondary lactase deficiency
  - Other lactose intolerance
  - Lactose intolerance, unspecified

Disorders of lipid absorption and transport
- Pancreatic colipase deficiency
- Pancreatic triacylglycerol lipase deficiency

Disorders of vitamin and non-protein cofactor absorption and transport
- Disorders of cobalamin metabolism and transport
  - Methylmalonic aciduria, vitamin B12 responsive
    - Vitamin B12 responsive methylmalonic aciduria type cbl A
    - Vitamin B12 responsive methylmalonic aciduria type cbl B
    - Vitamin B12 responsive methylmalonic aciduria type cbl mut-
  - Haptocorrin deficiency
  - Other genetic defects of cobalamine transport and metabolism
  - Secondary non-genetic disorders of cobalamin absoprtion, transport and metabolism
  - Unspecified disorders of cobalamin absorption, transport and metabolism
  - Constitutional megaloblastic anaemias due to vitamin B12 metabolism disorder
    - Thiamine-responsive megaloblastic anaemia syndrome
    - Homocystinuria without methylmalonic aciduria
      - Methylcobalamin deficiency type cbl E
      - Methylcobalamin deficiency type cbl G
      - Methylcobalamin deficiency type cbl Dv1
    - Congenital megaloblastic anaemia due to transcobalamin II deficiency
    - Methylmalonic aciduria - homocystinuria
      - Methylmalonic aciduria - homocystinuria type cbl C
      - Methylmalonic aciduria - homocystinuria type cbl D
      - Methylmalonic aciduria - homocystinuria type cbl F
    - Vitamin B12 deficiency anaemia due to congenital intrinsic factor deficiency
    - Vitamin B12 deficiency anaemia due to selective vitamin B12 malabsorption with proteinuria
- Disorders of folate metabolism and transport
  - Formiminoglutamic aciduria
  - Hereditary folate malabsorption
  - Neurodegenerative syndrome due to cerebral folate transport deficiency
  - Homocystinuria due to methylenetetrahydrofolate reductase deficiency
  - Other genetic disorders of folate transport and metabolism
  - Secondary disorders of folate transport and metabolism
  - Non genetic cerebral folate deficiency due to antoantibodies
  - Unspecified disorders of folate transport and metabolism
- Disorders of vitamin D metabolism and transport
  - Hypocalcaemic rickets
    - Hypocalcaemic vitamin D-dependent rickets
    - Hypocalcaemic vitamin D-resistant rickets
  - Hypophosphataemic rickets
    - Autosomal dominant hypophosphataemic rickets
    - Autosomal recessive hypophosphataemic rickets
    - Hypophosphataemic rickets with hypercalciuria
    - X-linked hypophosphataemia
- Disorders of metabolism and transport of other vitamins and cofactors
  - Biotin-responsive basal ganglia disease
  - Friedreich-like ataxia with selective vitamin E deficiency
  - Hereditary hypercarotenaemia and vitamin A deficiency
  - Hereditary vitamin K-dependent coagulation factors deficiency
    - Hereditary factor II [prothrombin] deficiency
      - Dysprothrombinaemia
      - Hypoprothrombinaemia
    - Hereditary factor VII [proconvertin] deficiency
    - Hereditary factor X [Stuart-Prower factor] deficiency
    - Combined deficiency of vitamin K-dependent clotting factors
  - Pantothenate-kinase-associated neurodegeneration
  - Pyridoxal phosphate-responsive seizures
  - Retinol binding protein deficiency
  - Sulfite oxidase deficiency due to molybdenum cofactor deficiency
    - Molybdenum cofactor deficiency, complementation group A
    - Molybdenum cofactor deficiency, complementation group B
    - Molybdenum cofactor deficiency, complementation group C
  - Thiamine-responsive megaloblastic anaemia syndrome
  - Multiple carboxylase deficiency
    - Multiple carboxylase deficiency due to biotinidase deficiency
    - Multiple carboxylase deficiency due to holocarboxylase synthetase deficiency
  - Microcephaly, Amish type

Disorders of mineral absorption and transport
- Disorders of copper metabolism
  - Benign familial copper deficiency
  - Wilson disease
  - Menkes disease
  - X-linked cutis laxa
- Disorders of iron metabolism
  - Iron overload diseases
    - Hereditary haemochromatosis
      - HFE-related hereditary haemochromatosis, Type 1
        - Arthropathy in haemochromatosis
      - Juvenile hereditary haemochromatosis, Type 2
        - Juvenile hereditary haemochromatosis, Type 2A
        - Juvenile hereditary haemochromatosis, Type 2B
      - Non-HFE-related hereditary haemochromatosis, Type 3
      - Non-HFE-related hereditary haemochromatosis, Type 4
    - Friedreich ataxia
      - Hereditary Optic Neuropathy associated with hereditary ataxias
    - Secondary iron overload
      - African iron overload
    - Atransferrinaemia
    - Microcytic anaemia with liver iron overload
    - Neurodegeneration with brain iron accumulation
      - HARP syndrome
      - Infantile neuroaxonal dystrophy
      - Neurodegeneration with brain iron accumulation due to C19orf12 mutation
      - Pantothenate-kinase-associated neurodegeneration
        - Atypical pantothenate kinase associated neurodegeneration
        - Classic pantothenate kinase associated neurodegeneration
      - Acaeruloplasminaemia
- Disorders of zinc metabolism
  - Hyperzincaemia and hypercalprotectinaemia

- Acrodermatitis enteropathica
- Acquired zinc deficiency syndrome
- Disorders of phosphorus metabolism and phosphatases
- Disorders of magnesium metabolism
  - Hypermagnesaemia
  - Hypomagnesaemia
    - Primary familial hypomagnesaemia
      - Familial hypokalaemia - hypercalciuria
      - Familial hypokalaemia - hypomagnesaemia
      - Familial hypomagnesaemia - hypercalciuria - nephrocalcinosis - severe ocular involvement
      - Familial hypomagnesaemia - hypercalciuria - nephrocalcinosis
      - Glaudemans type isolated autosomal dominant hypomagnesaemia
      - Hypomagnesaemia caused by selective magnesium malabsorption
      - Hypomagnesaemia with hypocalciuria
      - Hypomagnesaemia with normocalciuria
    - Secondary hypomagnesaemia
      - Drug-induced hypomagnesaemia
      - Neonatal hypomagnesaemia
        - Hypomagnesaemic tetany in newborn
  - Disorders of calcium metabolism
  - ~~Other disorders of mineral metabolism~~
  - ~~Disorder of mineral metabolism, unspecified~~

Disorders of fluid, electrolyte and acid-base balance
- Volume depletion
- Hyperosmolality and hypernatraemia
- Hypo-osmolality and hyponatraemia
- Acidosis
  - Acute respiratory acidosis
  - Anion gap metabolic acidosis
  - Chronic respiratory acidosis
  - Non-anion gap metabolic acidosis
- Alkalosis
  - Acute respiratory alkalosis
  - Chloride non-responsive metabolic alkalosis
  - Chloride responsive metabolic alkalosis
  - Chronic respiratory alkalosis
  - Mixed metabolic and respiratory alkalosis
- Mixed disorder of acid-base balance
- Hyperkalaemia
- Hypokalaemia
- Fluid overload
  - Fluid overload with generalised oedema
  - Fluid overload with localized oedema
  - Fluid overload with oedema
  - Fluid overload without oedema
- Other disorders of electrolyte and fluid balance, not elsewhere classified

Amyloidosis
- Beta-2-microglobulinic amyloidosis
- Familial amyloidosis, Finnish type
- Familial renal amyloidosis
- Primary amyloidosis
- Secondary amyloidosis
- Transthyretin-related amyloidosis
  - Familial amyloid polyneuropathy
  - Transthyretin-related familial amyloid cardiomyopathy
- Autonomic neuropathy due to amyloidosis
- Non-neuropathic heredofamilial amyloidosis
  - Cardiac non-neuropathic heredofamilial amyloidosis
  - Cerebral non-neuropathic heredofamilial amyloidosis angiopathy
  - Glomerular disorders in non-neuropathic heredofamilial amyloidosis
- Neuropathic heredofamilial amyloidosis
  - Cardiac neuropathic heredofamilial amyloidosis
  - Cerebral neuropathic heredofamilial amyloidosis angiopathy
  - Glomerular disorders in neuropathic heredofamilial amyloidosis
- Heredofamilial amyloidosis, unspecified
- Secondary systemic amyloidosis
  - Cardiac secondary systemic amyloidosis
  - Cerebral secondary systemic amyloidosis angiopathy
  - Glomerular disorders in secondary systemic amyloidosis
- Organ-limited amyloidosis
  - Cardiac organ-limited amyloidosis
  - Cerebral organ-limited amyloidosis angiopathy
  - Glomerular disorders in organ-limited amyloidosis
  - Amyloidosis of liver
  - Amyloidosis of small intestine
- Certain amyloidosis
  - Cardiac other amyloidosis
  - Cerebral other amyloidosis angiopathy
  - Glomerular disorders in other amyloidosis
  - paramyloidosis
- ~~Amyloidosis, unspecified~~
  - Cardiac amyloidosis, unspecified
  - Cerebral amyloid angiopathy, unspecified
  - Glomerular disorders in amyloidosis, unspecified
- Cutaneous amyloidosis
  - Primary (systemic) amyloidosis affecting skin and oral cavity
    - Macroglossia due to primary (systemic) amyloidosis
  - Primary localized cutaneous amyloidosis
    - Macular amyloidosis
    - Papular amyloidosis (Lichen amyloidosus)
    - Nodular amyloidosis
- Amyloid AL
- Amyloid AA
- Other amyloid and fibrillary deposition diseases
  - Monoclonal immunoglobulin deposition disease [MIDD]
  - Heavy chain deposition disease
  - Light and heavy chain deposition disease
  - Light chain deposition disease
  - Fibrillary glomerulonephritis
  - Immunotactoid glomerulonephritis

Certain specified metabolic disorders
- Disorders of plasma-protein metabolism, not elsewhere classified
- Tumour lysis syndrome
- ~~Other specified metabolic disorders~~
  - Benign symmetric lipomatosis
  - fatty degeneration
  - liposynovitis prepatellaris
  - steatosis
- ~~Metabolic disorder, unspecified~~
- Tophaceous gout

Postprocedural endocrine and metabolic disorders, not elsewhere classified
- Postprocedural hypothyroidism
- Postprocedural hypoinsulinaemia
- Postprocedural hypoparathyroidism

Postprocedural hypopituitarism
Postprocedural ovarian failure
Postprocedural testicular hypofunction
Postprocedural adrenocortical(-medullary) hypofunction
~~Other postprocedural endocrine and metabolic disorders~~
~~Postprocedural endocrine and metabolic disorder, unspecified~~
Nutritional and metabolic clinical manifestations